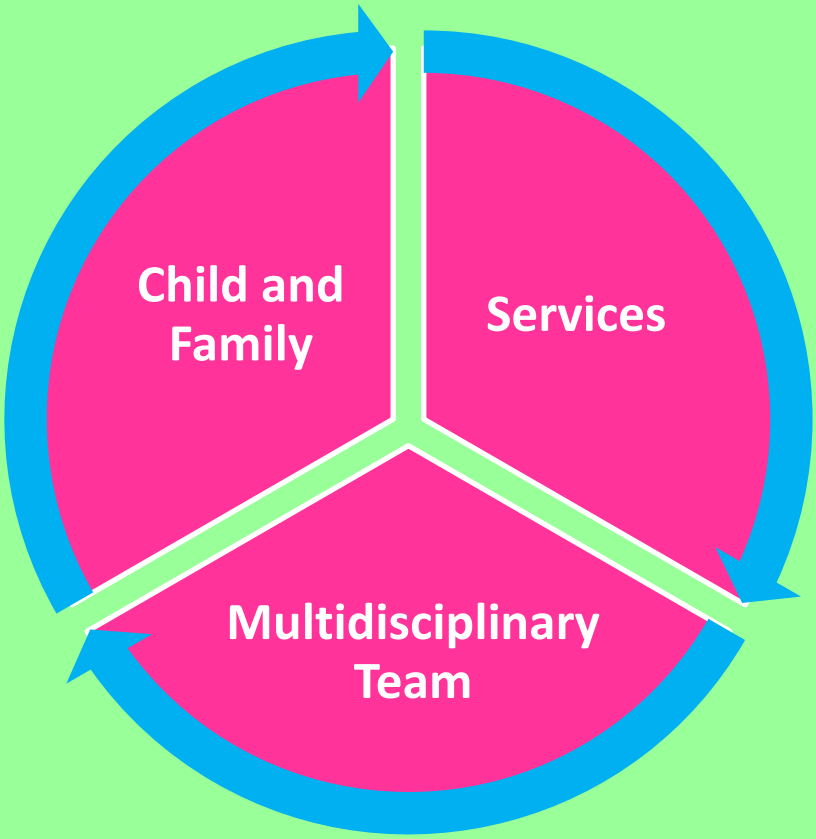

Impairment and Disability PY4017

Developmental Disability:

Service Delivery for individuals with Down Syndrome and Duchennes Muscular Dystrophy

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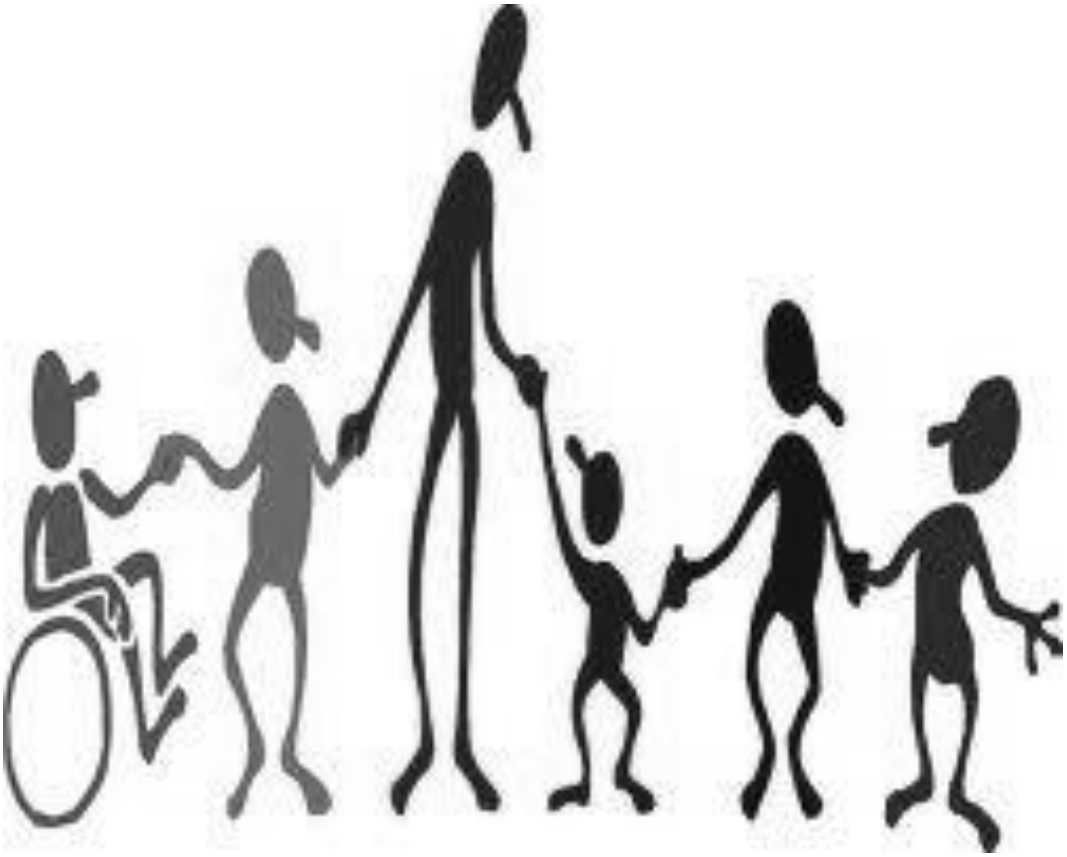
1. Services



1.1. Legislation

1.1.1 The National Disability Strategy 2004

The Government launched the National Disability Strategy as a framework of positive action measures for improving services for people with disabilities, and encouraging their participation in society. People with disabilities have the same legal, political, social and

economic rights as others; however, there may be difficulties in having those rights fulfilled. The National Disability Strategy, in conjunction with the Disability Act 2005 and the Citizens Information Act 2007, aims to provide new rights for people with disabilities and to establish a co-ordinated approach to services for people with disabilities.

The National Disability Strategy includes:

- The Education for Persons with Special Needs Act 2004 (EPSEN Act)
- The Disability Act 2005
- The Citizens Information Act 2007
- Six disability action plans that set out services to be provided to people with disabilities
- An investment programme for disability support services that runs over a number of years

Two pieces of legislation (see below) form a fundamental component of the National Disability Strategy. Both Acts set out a new approach to assessing the needs of eligible persons with disabilities and / or special educational needs for health and / or educational services:

- The EPSEN Act 2004 (http://www.oireachtas.ie/documents/bills28/acts/2004/A3004.pdf)
- Disability Act 2005 (http://www.oireachtas.ie/documents/bills28/acts/2005/a1405.pdf)

In addition, The Citizen's Information Act 2007 (http://www.oireachtas.ie/documents/bills28/acts/2007/a207.pdf) provides for the introduction of advocacy services for people with disabilities (Over 18s eligible).

The Disability Act 2005

The legal definition of disability in the Disability Act 2005 is:

"A substantial restriction in the capacity of the person to carry on a profession, business or occupation in the State or to participate in social or cultural life in the State by reason of an enduring physical, sensory, mental health or intellectual impairment."

The Act also states people are entitled to services if their disability is permanent (or likely to be permanent), results in significant difficulty in communication, learning or mobility or in significantly disordered cognitive processes and requires that services be provided continually to them.
For children with disabilities, services should be provided in early life to ameliorate their disability (Citizen's Information Board 2010).

The Disability Act 2005 provides for various measures including:

- Independent needs assessment for people with disabilities
- Independent redress and enforcement mechanisms,
- Obligations on public bodies to consider and respond to the needs of people with disabilities
- Six sectorial plans in the key service areas
- Targets for the employment of people with disabilities in the public sector.

Assessment of Need (AON) and Service Statement

- The Disability Act 2005 provides for the assessment of need of people with disabilities and the consequent drawing up of Service Statements.
- The purpose of an assessment is to decide what health and education needs arise from your child's disability and what services they require to meet those needs.
- Health services include personal social services and include services provided directly by the HSE and services provided on behalf of the HSE (many of the services for people with intellectual disabilities are provided by voluntary bodies on behalf of the HSE).
- The assessment of need highlights the range of health and educational needs associated with your child's disability and the services your child needs.
- It does not take into consideration the cost of the services or the State's capacity to provide them.
- The assessment of needs report is forwarded to a Liaison Officer who produces a service statement outlining the services and supports that will be provided.
- It is important to note that, due to financial and other constraints, it may not always be possible for your child to get all the services identified by the Assessment Officer in the Assessment Report.

Legislation regarding Education

- See Section 1.6. Education for details on the EPSEN Act 2004.

1.2 Family Centred Care

1.2.1 Principles of FCC

- It is a set of values, attitudes and approaches to services for children with special needs and their families.
- Family is the constant in the child's life and, the expert on the child's abilities and needs.
- The family works with service providers to make informed decisions about the services and supports that the child and family receive
- Strengths and needs of all family members are considered
- Each family should have the opportunity to decide the level of involvement they wish in decision-making for their child.
- The involvement of all family members should be encouraged.

(Law *et al* 2003) (CanChild Centre for Childhood Disability Research)

Premises (basic assumptions)

- Parents know their children best and want the best for their children.
- Families are different and unique.
- Optimal child functioning occurs within a supportive family and community context: the child is affected by the stress and coping of family members.

Guiding Principles ("should" statements)

- Each family should have the opportunity to decide the level of involvement they wish in decision-making for their child.

- Parents should have ultimate responsibility for the care of their children.
- Each family and family member should be treated with respect (as individuals).
- The needs of all family members should be considered.
- The involvement of all family members should be supported and encouraged.

(Rosenbaum *et al* 1998)

Table 1.2.1 Elements (key service provider behaviours)

1.2.2 Family Centred Services

A family centred approach is recommended in all services, by the professional literature as well as indicated by parent's needs and experiences. Parents may reject services and fail to benefit from professional expertise because professionals express a desire to control,

IMPORTANT FOR PHYSIOTHERAPY		
• To encourage parent decision making • To assist in identifying strengths and needs • To provide information and accessible services • To share information about the child and collaborate with parents	• To listen, respect and support families • To accept diversity • To believe and trust parents • To communicate clearly • To provide individualized services	• To consider psychosocial needs of all members • To encourage participation of all members and the use of community supports • To respect coping styles • To build on strengths

persuade, rescue, or prove professional capacities. Many services and professionals could benefit from reviewing the delivery of their service against the family centred services criteria.

The range of professionals that will provide services to children with developmental disability and their families is considerable. It includes: doctors, nurses, teachers, psychologists, speech and language therapists, physiotherapists, occupational therapists, social workers and respite care staff. The amount of training in counselling skills, listening skills, understanding family and child needs, and working with families that is included in their basic training will vary from substantial to very little or none. Some professionals training will have focused entirely on expert knowledge and skills. As already stressed, this is an issue for professional training bodies to address, as many young (and not so young) professionals have little preparation for working in a family centred way.

Basic guidelines to help health professionals improve the quality of their service, based on research studies and many years of working with families:

- Attitudes and beliefs - each individual believes the approach is the right one; they will have difficulty in changing their behaviour and practice.
- Listen to parents - parents are often the experts on Down syndrome and their knowledge of research and best practice may be more up-to-date than yours
- Demands of services - Recognise the demands that services and therapies may impose on families and try to develop family friendly models of delivery.
- The needs of fathers –The importance of fathers' full participation in decision making and in supporting his spouse should not be underestimated. It is a vital aspect for family well-being and for the child's progress.
- Share skills with parents – It is important to work with parents and pass on knowledge and practical skills to them as they are the child's best therapist and are with them every day.
- Co-ordinating services – It is important to co-ordinate services, to prevent families from becoming overwhelmed with too many demands and even conflicting demands.

(DSEI 2012)

FCS has been shown to:

- Empower parents,
- Improve emotional wellbeing – reduce stress, distress and depression
- Facilitate more positive perceptions of the child
- Improve parental satisfaction with services

(Can Child 2012)

1.2.3 Evidence behind Family Centred Service

- Studies have shown that parents and service providers highly value a family-centred approach to service provision (King *et al* 2000).
- Evidence from research studies on family-centred service can help to clarify what the benefits are to using family-centred service. Such information will give support to

recommendations for a more family-centred way of providing service to children and families.

1.2.4 Types of outcomes in a family-centred approach

- The purpose of working with families who have a child with special needs is to enhance the quality of life for all members (Fewell & Vadasy 1987). Consequently, the outcomes of interest for a family-centred approach have focused on more than just the child.
- A lot of the research on quality care has focused on the key outcomes of satisfaction with services, reduced stress and worry, and follow-through with therapy programs (King *et al* 1996).
- In addition, outcomes about siblings, the family, health care providers, the community, and the service organization have been considered in showing the benefits of family-centred service (Allen 1987; Bennett & Guralnick 1991; Epstein *et al* 1989).

1.2.5 Nature of the evidence?

- Do not demonstrate cause and effect.
- Some focus on one aspect of FCS whilst others focus on evaluating FCS as a whole.
- Children's rehabilitation or health care that is community-based – no in-hospital services.

1.3. Disability Services

1.3.1 Structure of HSE

Four areas:

- Each area is responsible for arranging for the provision of services to people in its area.
- Each area has a Disability Services Manager to organise service delivery to people with disabilities.
- HSE or voluntary organisations provide disabilities teams in some areas.

Entry to Services:

- If diagnosis is at time of birth, the hospital may refer you to appropriate services and supports.
- The Paediatrician or Disability Services Manager in your Local Health Office may refer your child to EI services.
- The parent can also directly apply to service providers or apply for AON under the Disability Act 2005.

1.4. Support and Psychological Services

1.4.1 Parent support services:

Down Syndrome Ireland - www.downsyndrome.ie/

- This organisation is run by people with DS and parents of children with DS
- There are 25 branches throughout Ireland
- The website provides a lot of information for new and expectant parents
- It provides a counselling service and information to parents and guardians of persons with Down syndrome, especially those with new-born babies.
- It keeps parents informed of developments taking place at home and abroad for the advancement and care of people with Down syndrome.
- It provides a forum where parents and guardians can meet and exchange views and ideas.

Muscular Dystrophy Ireland (MDI) - http://www.mdi.ie/

- Provide information and support to people with neuromuscular conditions and their families.
- Supports advocating for services to allow people with neuromuscular conditions to fully participate in society and live a life of their own choosing
- More details available through the above link.

Disabiltyinfo.ie

- This site gives information of services in different areas of Ireland: www.disabilityinfo.ie/index.php?page=18&lang=EN

1.5. Early Intervention Services

1.5.1 Early Intervention – What is it?

Early intervention programmes are specially designed to meet the specific needs of children with learning difficulties and disabilities. These programmes provide healthcare, education and treatments such as physiotherapy and speech and language therapy (SALT), as well as advice and support to the family looking after the child (HSE 2012).

The early intervention team focus on providing support to babies and children with Down Syndrome (DS), from when they are born until five years of age. Early intervention is important because the earlier a child with DS receives the help and support necessary, the healthier and more independent they are likely to become later in life (HSE 2012).

1.5.2 MDT in early intervention for DS

SALT – DS is associated with difficulties learning to speak. SALT intervention can help improve communication (HSE 2012).

Occupational Therapist (OT) – As a result of hypotonia, people with DS can have problems with tasks that involve physical co-ordination (feeding, dressing). An OT can break down tasks into small steps, which helps a person with DS learn how to complete the tasks step-by-step. OT's provide practical support to enable greater independence (HSE 2012).

Physiotherapist – At the early intervention stage physiotherapy focuses on facilitating motor control and co-ordination in order to achieve developmental milestones. Physiotherapists can help children with DS learn to roll over, sit up, and walk. They can help with range of movement (ROM) also (Galli *et al* 2008; HSE 2012).

1.6. Education: Supports and Services for Children with Disability and/or Special Educational Needs

The following information has been sourced from:

1. National Council for Special Education (NCSE) website - *Children with Special Educational Needs: Information Booklet for Parents (2011)* http://www.ncse.ie/for_parents/Information_Pamphlets.asp
2. Citizens Information website – *Entitlements for Children with Disabilities: http://www.citizensinformationboard.ie/publications/entitlements/downloads/children_with_disabilities.pdf*
3. Interviews with a Paediatric Physiotherapist *Mary Delaney* (SPRAOI Centre, Portlaoise), North Tipperary SENO *Carmel Callery*, Resource Teacher *Michelle McNulty*, and School Principals *Mary Gilligan* and *Tony Slattery*.

1.6.1 Introduction

The EPSEN Act 2004 explains special educational needs as "a restriction in the capacity of a person to participate in and benefit from education on account of an enduring physical, sensory, mental health or learning disability, or any other condition which results in a person learning differently from a person without that condition." It is recommended that children with special educational needs go to school "in an inclusive environment with children who do not have special educational needs" except if it is not in the best interests of the child or the other children in the class.

Under the EPSEN Act 2004, all children with special educational needs have the right to an education which corresponds to their needs. The aims of education for these children are the same as apply to all children:

"To enable children to live full and independent lives so that they can contribute to the community, cooperate with other people and continue to learn throughout their lives"

Special educational needs may arise from four different areas of disability:

- Physical
- Sensory
- Mental health
- Learning disability
- Or from any condition which results in the child learning differently from a child without the condition.

Wherever possible, children with special educational needs should be educated in an inclusive environment. However, there are some exceptions to this demand:

- If it is not in the best interest of the child, e.g. the learning environment is not suitable to the child's needs
- If it is not in the best interest of the other children with whom the child is to be educated, e.g. the child is hindering the education of other pupils

Types of Education Provision

Setting	**Definition**
Mainstream Class	A mainstream class in a mainstream primary or secondary school where the class or subject teacher has primary responsibility for the progress of all pupils in the class, including pupils with special educational needs. Additional teaching support from a learning/resource teacher or SNA may be provided, where appropriate.
Special Class (Mainstream)	A special class in a mainstream primary or secondary school with a lower pupil-teacher ratio specified according to category of disability. This means that classes have small numbers of pupils where more attention can be given to their particular requirements.
Special School	A special school with a lower pupil-teacher ratio specified according to category of disability. This means that classes have small numbers of pupils. For example a special school for children with moderate general learning disability has one teacher for every eight pupils. Similarly, SNA support is available to assist students with their care needs

throughout the school day

(NCSE 2011)

More detailed information on each of these educational settings can be found through the NCSE link provided at the start of this section.

1.6.2 Entry Process to Education

Parents are often very concerned about whether it is best for their child with special educational needs to attend a mainstream school, special class (mainstream) or a special school. In making this decision, parents are recommended to think about the following factors:

- the child's needs
- the child's best interests
- parents preference
- the needs of other children in the school setting

1.6.3 Who is Involved in the Decision of where the Child is to be Educated?

Parents:

As stated above, the decision of where a child is to be educated ultimately rests with the child's parents. However, parents of children with educational needs are recommended to seek advice and guidance from the following professionals as well as from other parents of children with disabilities or educational needs.

Special Educational Needs Organisers (SENO):

SENO's are employed by the state to provide a direct service to the parents of children with special educational needs. Integral to this service is helping parents to make informed decisions as to where their child may be best educated based on his/her needs. They do this by providing information about the various different schools available within their designated geographical area. A key role of the SENO is to ensure that the chosen school is sufficiently resourced to cater for the needs of a child with disability. While they are not involved in the assessment of the child or the school environment, they are the link for parents, schools and

clinicians to the state. SENO's can request resources from the state to support the child both physically and educationally in the chosen establishment.

The School:
The principal and teachers within a school often know most about their school environment. Therefore they are a key point of contact. Once the child commences his/her education within a school, the teacher and/or principal should be able to quickly identify whether the chosen environment is suitable for the child. If it is not, further resources may be required for the child.

Educational Psychologist:
The educational psychologist will complete a comprehensive educational assessment of the child. This may be carried out by a psychologist within the early intervention team or a National Educational Psychological Service (NEPS) psychologist if the child is enrolled in a school. By doing this assessment, the child's needs and abilities can be established which can help guide parents with the decision making process. It also allows the school to resource itself to meet the child's needs prior his/her starting in a school.

Early Intervention Team (EIT):
The EIT is a vital source of information for parents deciding where to send their child. This information is usually available in the AON report which is described later. For children who have physical impairments, it is important to consider how the child ambulates around the school and the level of assistance required. The OT and/or physiotherapist should assess the child in the school environment so that adjustments can be made to promote independence.

1.6.4 How is a Child's Educational Needs Assessed?

Early Years:
In most cases the key to providing a good start to children with educational needs in school education is through early intervention (EI). Under the Disability Act (2005), all children with a diagnosed disability under the age of 5 are entitled to an EI assessment. An Assessment of Need (AON) can be drawn up which includes an assessment of the child's educational needs. The AON can then be presented to the chosen school before the child

commences their education. This helps optimise the child's learning as resource structures can be put in place prior to the commencement of school.

At School Going Age:
For children with educational needs, their delayed learning may not become evident until they start school. Often it is the teacher of the student who first identifies the child's educational needs. Parents, teachers and principals can then discuss the matter and bring it to the attention of the NEPS psychologist. Based on this assessment, the child can then be granted resource teaching through the SENO to help facilitate his/her learning. The NEPS psychologist may refer the child to other services such as speech and language if they feel the child requires further management. Furthermore, NEPS has developed a system of identifying and supporting children in the school environment, known as the Continuum of Support. More information can be found through the NCSE website.

1.6.5 Support Services Available During the School Lifespan

To optimise the child's ability to participate in school life and access the curriculum, the Department of Education and Skills (DES) and the Health Service Executive (HSE) have developed a number of services and systems to support the child. These services support children through pre-school, primary school, and secondary school. A list of the state bodies and agencies responsible for the education and support of children can be found through the NCSE website (see start of section).

Early Childhood Care and Education (ECCE):
Under the Irish Constitution, all children are required to commence education by the age of six. However, before this age, children can avail of early childhood education. Since January 2010, the ECCE Scheme provides a free year of early childhood care and education for all children aged between 3 years 2 months and 4 years 7 months, regardless of family income or ability to pay. Education professionals report that children with special educational needs who avail of this pre-school year have a better ability to adjust and adapt to the structure of school life than those who don't.

Children's Services Team:
The Children Services Team is a coalition of the HSE and Enable Ireland. They are an MDT whose purpose is to provide health and therapeutic support to children with disabilities

throughout their school life. The overall aim of the service is to enhance the child's ability to be educated and to foster independence in the child.

The Children's Services Team consists of:

1. Early Intervention Team (EIT 0-5yrs)
2. School Aged Team (SAT 6-18yrs)

Members include:

- Childcare Staff
- Consultant Paediatrician
- Occupational Therapist
- Physiotherapist
- Social Worker
- Psychologist
- Speech and Language Therapist

From the age of 0-5, the Children's Services Team support children through EI which can be provided to children in various different settings. Once a child enters education, they are supported by the SAT. Children with disabilities who have multiple areas of need are managed by individual members of the SAT as required throughout their school life. The SAT provides on-going assessment, monitoring and treatment. A child with disability who has only one area of need often will not require MDT management and thus may not be managed by the SAT. These children are often referred onto and managed by the community primary care team.

Members of the Children's Services Team may be directly involved with the child's school life and work in conjunction with the teachers to enhance their capacity to be educated in the best way possible. This is often the situation for children with disabilities who are attending special schools.

Resource and Learning Support Teachers:
Resource hours and learning support are systems put in place to support children with special educational needs. These educational resources are allocated every school in two main ways:

1. The General Allocation Model (GAM)
2. Through application to the NCSE by work of the SENO

In general, learning support is granted to those children who have milder learning needs. Children are taught purely academic skills during learning support hours. Hours are distributed by the school under the GAM. Resource hours are granted to children with significant physical difficulties or more profound learning needs and are distributed by the SENO.
Under the GAM, additional learning support hours are granted to schools based on their enrolment numbers and are used to cater for children with mild speech, coordination or learning difficulties (high incidence special educational needs).

High Incidence Needs = those which occur more frequently in the general population, e.g. mild general learning disability

Low Incidence Needs = those which occur less frequently in the general population, e.g. severe general learning disability and autism.

The NCSE, through its SENO network, grants resource support to schools for children with low incidence special educational needs. Presently, the maximum number of resource hours which can be allocated to a school to support a child is 5 hours per week. Unlike learning

support, resource hours are not for purely academic use. During this time, the resource teacher (or other) can use the time to help promote the child's independence. For example, this may be teaching a child with a physical impairment to take on and off his/her shoes or helping a child with speech impairment with his/her phonetics.

Table 1.6.2 How Learning Support and Resource Hours are distributed

<table>
<tr><td rowspan="3">Learning Support</td><td colspan="6">General Allocation Model (GAM)</td></tr>
<tr><td rowspan="2">Children who score below 10th Percentile on Standardised Testing</td><td colspan="5">Special Educational Needs (High Incidence)</td></tr>
<tr><td colspan="2">Borderline-Mild GLD</td><td>Mild GLD</td><td colspan="2">Specific Learning Disability</td></tr>
<tr><td rowspan="3">Resource Teaching</td><td colspan="6">NCSE (SENO)</td></tr>
<tr><td colspan="6">Special Educational Needs (Low Incidence)</td></tr>
<tr><td>Moderate-Severe Learning Disability</td><td>Significant physical or sensory impairment</td><td>Emotional disturbance and/or behavioural problems</td><td>Autistic Spectrum Disorder</td><td>Speech and Language Difficulty</td><td>Coordination or Attention Difficulty (ADHD, Dyspraxia)</td></tr>
</table>

*GLD = General Learning Disability (NCSE 2011)

More information of Learning Support and Resource Teaching can be found on the NCSE website (link provided at the start of section).

Emergency Works:
For children with physical disabilities, mobilising around a school can be quite challenging, and sometimes not possible. The emergency works scheme allows children with physical disability to independently mobilise about the school establishment. Grants are available for children entering a school which is not equipped to cater for their physical handicaps or for those children who are current students but their physical disabilities are deteriorating. Such grants are used to construct ramps, rails, chair lifts, elevators and special seating.

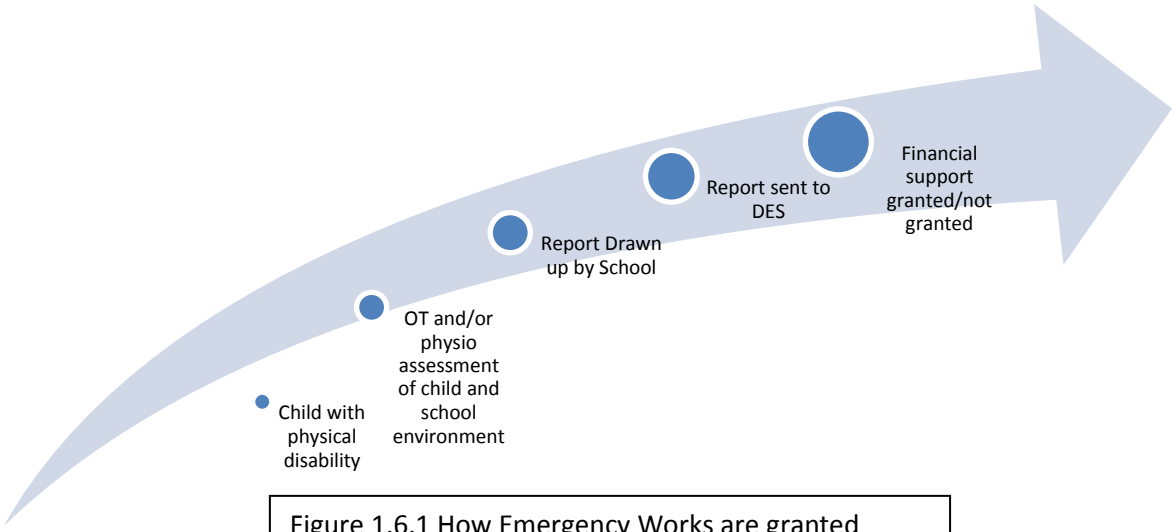


Figure 1.6.1 How Emergency Works are granted

Special Needs Assistant (SNA):

The NCSE provides SNA's to support children with special care needs in mainstream schools. The work of the SNA includes helping with activities such as clothing, feeding, and toileting. They are strictly not involved in the education of the child. While the child's ability to function within the school and the classroom is enhanced through the SNA, the SNA must promote the child's independence at all times. Their role is therefore a facilitating one in which they assist rather than problem-solve for the child. This is to help children develop and acquire independent living skills they will need in the future. SNA support is granted by the school through the SENO who must justify how and why SNA support will benefit the child. A report from the health professional who diagnosed the care needs of the child is also required. The following are the criteria under which a SNA may be requested:

- The child has a significant medical need for care assistance
- The child has a significant impairment of physical or sensory function, or
- The child's behaviour is such that they are a danger to themselves, other pupils, or that it significantly interferes with the learning opportunities of their peers.

Assistive Technology:

Assistive technology is available to promote the education of children with special educational needs and/or physical disabilities. Grants for assistive technology such as radio aids, Braille machines and computers are provided to support interactive learning and to enable the child to access the curriculum. Further information on the criteria required to be

granted such equipment can be found through the following link:
http://www.sess.ie/sites/default/files/Circular_M14_05.pdf

Visiting Teachers:
Students who have a disability which is significant enough for them to lose or seriously hinder their vision or hearing can avail of the visiting teacher scheme. The visiting teacher helps devise the optimal method for the student to give and receive information an inclusive setting so that they can communicate and learn optimally. The roles of the visiting teacher can be found on the NCSE website (link provided above).

Transport:
If parents decide to send their child to the nearest recognised mainstream/special school that is or can be resourced, the DES will provide a mode of transport to meet the child's special educational needs. In certain circumstances the Department will sanction grant-aid towards the cost of private transport. In some cases, parents can avail of 'escorts' who will safely bring their children with special educational needs to and from the chosen school. It is often the SENO who organises transport support for children with disabilities.

Child and Adolescent Mental Health Services (CAHMS):
CAHMS is a service which supports children up to and including the age of 16 with on-going mental health problems that affect their day-to-day living and relationships, impacting on their school education. These problems are commonly referred to as an emotional and behavioural disorder (EBD).

Children who are referred to CAHMS often present with one or more of the following:

- Anxiety or worries
- Depression and low mood
- Behavioural difficulties
- Developmental difficulties
- Attention Deficit Hyperactivity Disorder (ADHD)
- Psychosis, i.e. "losing touch with reality"
- Self-harm
- Eating problems including anorexia nervosa and bulimia
- Family relationship difficulties

(Second Annual CAMHS Report 2009-10)

CAHMS is strictly uninvolved in the education of children. The service offers assessment, intervention and therapeutic support to both the child and his/her family. Mental health problems may often first become apparent to the child's teacher and/or SNA who are obliged to report any concerns they have to the principal. Discussion with parents and further investigation may then be required. Further information can be found through the following link:

http://www.hse.ie/eng/services/Publications/services/Mentalhealth/camhs2010.pdf

1.6.6 Transition through School Life

Transition is an inevitable feature of school life for every child. These transitions are:

- Pre-school to primary
- Primary to post-primary (secondary)
- Post-primary to higher education or adult life

Coping with the various changes involved at each transition is a challenge for every child going through the education process. For the child with disability this challenge is often accentuated. Planning is therefore paramount. Children with disabilities may sometimes transition between schools at various stages of their school life depending on their individual needs and abilities. More information can be found through the link at the start of the section.

(NCSE 2011)

1.6.7 Flaws in the Service Provision

- Funding - The NCSE report in 2011 expresses the growing difficulty for the organisation to maintain service provision to schools despite cuts in staffing numbers and funding.
- EPSEN Act only partly enacted - The NCSE has published a plan for the phased implementation of the EPSEN Act including 'Individual Education Plans' for each child assessed as having a special educational need. However this has not yet been implemented.
- Lack of communication and understanding between health and education professional

1.6.8 Third Level Education

- Application through Central Applications Office (CAO)

- Some institutions keep a number of places for individuals with disability.
- The Disability Liaison Officer in the third level institution can help provide information on applying and information on the college experience.
- The DARE (Disability Access Route to Education) scheme launched in October 2009 helps enable people with disabilities to attend third level education.
    - In 2010 there was 11 higher education institutions involved in the DARE scheme
    - Each institution involved has a number of places on a reduced CAO points basis for students qualifying through DARE
- There are a number of academic, personal and social supports offered to students with a disability while studying at third level including:
    - An orientation programme
    - Extra tuition if required and exam support
    - Access to assistive technology and training
    - Course materials in suitable format (e.g. audio tapes of journal articles)
- Student Grants: Students with disabilities may qualify for grants similarly as other students may. Also there are some grants specifically for people with disabilities.
- The Fund for Students with Disabilities by the Department of Education and Skills provides funding to higher education institutions for the provision of services and supports to full-time students with disabilities. The Fund helps ensure students with disabilities can participate fully and are not hindered by a disability.
- AHEAD (Association for Higher Education Access and Disability) is a voluntary organisation working to allow individuals with disabilities better access to third level education. See 'Accessing Third-level Education in Ireland –A Guide for Student with Disabilities' for further information and www.ahead.ie
- Other educational schemes:
    - Back to Education Allowance
    - Vocational Training Opportunities Scheme (VTOS)

1.7. Leisure and Sport Participation Services

1.7.1 Special Olympics (NB for DS)

'Special Olympics Ireland is first and foremost a sport organisation for people with an intellectual disability, but it provides athletes with far more than the physical benefits of sport. It's about fun, friendships and team spirit; it's about a feeling of belonging, and ultimately improving quality of life. Special Olympics changes lives in so many different ways. Through sport, athletes develop both physically and emotionally, they make new friends, realise their dreams, and know they can fit in. Special Olympics enable them to achieve and win not only in sport but in life too.'

(Special Olympics Ireland)

- People of all abilities are facilitated to participate at local, regional and national level. The emphasis is on participation and inclusion not winning.
- There are branches throughout Ireland providing the opportunity to participate in 15 different sports.
- People must be 8 years old to compete but they can begin training at 2.5 years.
- Having divisions in competitions ensures that individuals are competing against similar ability. This is based on age, sex and previous performance in competitions or training.
- To prevent individuals taking advantage of this system the Maximum Effort rule was introduced. This prevents individuals under-performing in preliminary rounds so that they will be placed in lower divisions and have a higher probability of winning the final. This rule outlines that any athlete who is 15 % better that their entry score is disqualified.
- 1^{st} to 8^{th} place is rewarded, with medals for 1^{st} 3 places, and 4^{th}-8^{th} place with ribbons. There are also rewards for significant accomplishments.
- They also have a programme for unified sports to promote an inclusive participation between those with and without a disability. The abilities within the teams can differ but must be similar to those that they are competing against.

Rules for participating are available at http://media.specialolympics.org/soi/files/resources/Sports-Rules-Competitions/2012RulesChange/2012Article1.pdf

For more information: www.specialolympics.ie

Other services in Ireland for children with disability to enhance participation

IWA-Sport

- The national governing body in Ireland for sport for people with physical disability
- Aims to develop and promote opportunities for people with a physical disability to participate in sport at their level of choice.
- Sports on offer are athletics (track and field), archery, basketball, bowls, snooker, pool, table tennis, boccia, badminton, tennis, wheelchair rugby, swimming.
- IWA-Sport has sport and leisure programmes to suit all ages.
- Website: www.iwasport.com

Riding for the Disabled Association Ireland (RDAI)

- Aims to provide riding and driving opportunities for people with a mental or physical disability.
- Email: rdaise@eircom.net

Irish Disabled Sailing Association

- Organisation which promotes the participation of people with disabilities in the sport of sailing.
- Website: www.sailforce.ie

1.7.2 Guidelines to improve PA in youths (With and without a disability)

http://health.gov/paguidelines/midcourse/PAG_Mid-course_Report.pdf

http://health.gov/paguidelines/guidelines/default.aspx#toc

1.7.3 PA in different conditions:

http://www.ncpad.org/DisabilityIndex/kids

1.7.4 Irelands PA guidelines:

http://www.getirelandactive.ie/content/wp-content/uploads/2011/12/Get-Ireland-Active-Guidelines-GIA.pdf

1.8. Supporting Transition from Paediatric to Adult Services:

1.8.1 Transition to adulthood and discharge from child services:

- Process should begin in adolescents by gradually giving a child more independence and responsibility in treatment.
- They become more involved in planning their education pathway and what they want to achieve.
- A transition plan should be put in place that recognises their individual strengths and weaknesses and any services that the child may need to help him/her realise their plans for adult-life.

(Van Cleve *et al* 2006)

Transition from paediatric to adult services can be a stressful time for young people with disabilities and their families. Therefore it is a time when families need significant access to support, advice and planning. The responsibility of the family in helping the transition is also important. If the individual with a disability is without strong parent advocates, they may receive a lower level of support during the transition, which may affect health service care, social, financial and further educational opportunities. In order to facilitate the transition, a key worker can act as an advocate and provide information and support to the young person and their family.

(Aniapravan *et al* 2012)

1.8.2 Duchenne Muscular Dystrophy (DMD)

- In DMD the transition between services is also a time when their condition can change significantly, therefore heightening the importance of a smooth bridge between services.
- There is currently a poor bridge between paediatric and adult services in the UK. A survey in the UK reviewing service provision for adults with chronic neuromuscular disorders highlighted concerns regarding the transition process and the availability of specialist adult services in certain areas of the UK. At present, there is much variability across the UK in service provision for the DMD patient group.

- Young people with neuromuscular disease express the need for access to further information on adult services and a key worker to facilitate the transition. They also feel that in current practice, the transition is too sudden and should be a gradual process.
- In a qualitative study by Abbot and Carpenter (2010) looking at the transition from child to adult services, all of the 37 men interviewed had completed full time education, were unemployed at home and had very restricted social lives. They were also heavily dependent on their family for personal care and to facilitate social interaction with others. A lack of transition planning emerged from the qualitative study and the majority of families described the overall experience of service provision as 'problematic'.

(Abbot 2012; Aniapravan *et al* 2012)

1.9. Vocational Services

(See www.citizensinformation.ie for further information)

1.9.1 Training & Employment

- People with disabilities can access two types of training:
    - Mainstream Vocational Training
    - Rehabilitative Training
- Sheltered Work
- Mainstream Employment

1.9.2 Training

- HSE provides guidance counsellors who offer information on rehabilitative training (focusing on improving core life skills, social skills and basic employment skills) and sheltered work services.
- Vocational training, primarily provided by FÁS, focuses on particular skills which help individuals with a disability to receive employment. See http://www.fas.ie/en/ for further information.

1.9.3 Sheltered Work

- Allows individuals with disabilities to take part in daily work in an environment where personal support services are provided. For further information see the Irish Association of Supported Employment (www.iase.ie)

1.9.4 Back to Work

- The individual with a disability may qualify for back to work supports if they are receiving Disability allowance. See www.citizensinformation.ie for further information.

1.9.5 Employment

- The Disability Act 2005 outlines legal obligations of public service bodies to support employment for individuals with disabilities. The Act states that 3% of employment for public service bodies is reserved for individuals with disabilities.
- There are a number of supports for people with disabilities in employment, including adaptations grants and disability awareness training for employers.
- Individuals with disabilities qualify for a number of 'back to work supports' including the Community Employment Scheme and the Community Services Programme.
- Details of employment related supports for people with disabilities can be found at http://www.citizensinformation.ie/en/employment/employment_and_disability/
- There are supports available for the self-employed also.

For more information contact your local FÁS office or go to www.citizensinformation.ie

1.10. Accommodation Services

(See www.citizensinformation.ie for further information)

- Accommodation options may include privately owned, privately rented or local authority housing depending on the nature of the disability and the financial situation
- Other options may include sheltered housing, community group homes and residential care.
- Various housing supports are available for people with disabilities.
- There are supports available for home adaptation if necessary to facilitate independent living.

- The individual with disability may be eligible for home help or a personal assistant.

1.10.1 Voluntary Housing Organisations

- Independent non-profit organisations that provide rented housing for older people, people with a disability or people who are unable to afford to purchase a home.
- Some voluntary organisations provide non-housing services, for example group meals, social activities and welfare advice.

1.10.2 Home Adaptations

- Support may be provided to make house adaptations so the house is more appropriate for the individual's needs.
- Financial aid can be accessed.
- To be eligible for financial aid, an OT assessment of the home may be required.
- Other health professionals, such as public health nurse or a physiotherapist can also provide advice on home adaptations.
- If on a low income there are other supplements and schemes in relation to housing, which are non-specific to disability.
- See http://www.citizensinformation.ie/en/housing/housing_grants_and_schemes/housing_adaptation_grant_for_people_with_disability.html for full details.

1.10.3 Accessibility

- All houses that were built in Ireland after 2001 were required to be "visitable" by people with disabilities:
    - Low-level entrance
    - Bathroom at living room level

1.10.4 Centres for Independent Living

- Formed by and for individuals with disabilities to encourage independent living and full participation in society.

1.11. Community Care Services

Full details on the following can be found at http://www.citizensinformationboard.ie/publications/providers/downloads/Entitlements_Disabilities.pdf

Services provided by the HSE to help people with disabilities to remain living in their communities, including:

- Home Nursing Services
- Home Help
- OT
- Social Work services
- Meals on wheels
- Personal Assistant Service
- Home Care Attendants

1.12. Advocacy Services

Aim to empower people with disabilities by informing them of their entitlements and when required, negotiating on their behalf. The Citizen's Information Board supports forty-seven advocacy projects around Ireland.

1.13. State Allowances & Entitlements

The following section covers entitlements available for children and adults with disabilities in Ireland. The information provided has been synthesized from the following reliable sources:

- http://www.hse.ie/eng/services/Find_a_Service/entitlements/
- http://www.citizensinformation.ie/en/

- http://www.downsyndrome.ie/index.php/entitlement
- www.welfare.ie

1.13.1 Citizen Information

- A nationwide network of Citizens Information Services (CISs) provides free, confidential and impartial information on all aspects of rights and entitlements.
- The Citizens Information website (www.citizensinformation.ie) gives detailed online information on all the benefits and entitlements.
- CIS are responsible for Money Advice and Budgeting Service and support advocacy services for individuals with disabilities.
- The Citizen's Information Board support people with disabilities in identifying their needs and choices.
- It is an excellent resource for families who have a child with disability.
- The Citizen's Information Board provide booklets on 'Entitlements for Children with Disabilities' and 'Entitlements for People with Disabilities', which are key sources of information for individuals living with disability.
    - http://www.citizensinformationboard.ie/publications/providers/downloads/Entitlements_Disabilities.pdf
    - http://www.citizensinformationboard.ie/publications/entitlements/downloads/children_with_disabilities.pdf

People, parents, carers and guardians of people living with a disability are entitled to some level of assistance from the Irish State. Entitlements are as follows:

1.13.2 Entitlements for people with a disability

- Disability Allowance
- Long Term Illness Scheme
- Medical/GP Visit Only Card

The HSE's complete guide to Medical and GP visit cards available at:http://www.hse.ie/eng/

1.13.3 Entitlements for Carers

- Domiciliary Care Allowance
- Carers Allowance
- Carers Benefit
- Respite Care Grant
- Employment Leave
- Carers Leave

There are also secondary benefits that carer's may be entitled to. People with disabilities on certain payments are automatically entitled to these allowances. See http://www.citizensinformation.ie/en/ for further details.

1.13.4 Payments for people with children may include:

- One-Parent Family Payment
- Child Benefit (Note Budget 2013 cuts to Child Benefit).
- Early childcare Supplement
- Family Income Supplement
- Supplementary Welfare Scheme
- Back to school clothing and footwear allowance

- See http://www.citizensinformationboard.ie/publications/providers/downloads/Entitlements_Disabilities.pdf for further info.

1.13.5 Tax Credits/Allowances

There are some extra tax credits and allowances which are available to people with disabilities and their carers. Generally the tax legislation uses the term "incapacity" rather than "disability". See further details on the tax revenue website.

- Incapacitated Child Tax Credit
- Home Carers Tax Credit
- Employing a carer Allowance

1.13.6 Aids and Appliances

- If you are a medical card holder, the HSE will help with the purchase costs of necessary medical and surgical aids and appliances (such as wheelchairs and walking aids).

- If you do not have a medical card, you may get some assistance from the HSE if you are unable to meet the costs from your own resources and the aids and appliances are considered necessary as part of hospital treatment.
- If you have one of the conditions designated under the Long-Term Illness Scheme, you may also get assistance towards the purchase costs of medical and surgical appliances for that condition.

1.13.7 Making Decisions on Medical Treatment

- You have the legal right to make decisions on medical treatment for your child up to the age of 18.
- Children aged 16 and over may be allowed to provide valid consent to some medical procedures.
- After the age of 18, you no longer are entitled to make decisions on your child's behalf.
- But in reality, parents of children with intellectual disabilities often make decisions on behalf of their adult child (Citizen Information Board 2010)

1.13.8 When the Caring Period Ends

- If the person you are caring for dies, Carer's Allowance or Benefit continues to be paid for six weeks after the death.
- You should notify the Department of Social and Family Affairs regarding the death as soon as possible.
- You may be eligible for a Bereavement Grant if PRSI conditions are met.

Useful Link for Families:

- www.informingfamilies.ie.

1.14. Paediatric Palliative Care Services

1.14.1 What is Palliative Care?

- According to WHO (2013) palliative care for children is the care of the child's "body, mind and spirit". Health care professionals must work to manage the child's physical, psychological, and social distress.
- It commences at initial diagnosis of illness and continues whether or not the child is receiving treatment for the disease.
- A multidisciplinary approach that includes the family (and involves giving them support) must be implemented for effective palliative care
- Palliative care can be provided in the child's home, tertiary care facilities and in community health centres.
- Children with life-limiting conditions should ideally be cared for by staff trained in paediatrics, and palliative care services should be accessible, flexible, equitable and appropriate to meet the needs of all children with a life-limiting condition and their families.

(DOHC 2010; WHO 2013)

1.14.2 Conditions Requiring Palliative Care

Table 1.14.1 Classifications of Paediatric Conditions Requiring Palliative Care

	Description	**Examples**
Group 1	Life-threatening conditions for which curative treatment may be feasible, but can fail. Where access to palliative care services may be necessary when treatment fails children in long term remission or following successful curative treatment are not included.	Cancer, irreversible organ failures of heart, liver, kidney.
Group 2	Conditions where premature death is inevitable, where there may be long periods of intensive treatment aimed at prolonging life and allowing participation in normal activities.	Cystic fibrosis
Group 3	Progressive conditions without curative treatment options, where treatment is exclusively palliative and may commonly extend over many years.	Batten disease, mucopolysaccharidoses, muscular dystrophy
Group 4	Irreversible, but non-progressive conditions causing severe disability leading to susceptibility to health complications and likelihood of premature death.	Severe cerebral palsy, multiple disabilities such as following brain or spinal cord insult

1.14.3 How does paediatric palliative care (PPC) differ from adult palliative care?

- The number of children dying is small, and their conditions are generally rare with diagnoses specific to childhood.
- Prognosis can be difficult to predict and the palliative phase can often be longer, and more episodic and unpredictable.
- The model of family-centred care is used as the care embraces the whole family.
- There is a heavy responsibility on the parents for personal and nursing care, so adequate resources to support them are necessary.
- Siblings are vulnerable and parents must provide continuous care for them as well as providing full time care to a sick child.
- Some conditions may run in families so other children may be living with, or have died from, the same condition.
- It is essential to provide education and play to the sick child.
- A child's ability to communicate and understand will vary greatly according to their age or stage of development.

(DOHC 2010)

1.14.4 What are the specific needs of adolescents in palliative care?

- Involvement in decision-making is important but it can often be a source of conflict if parents want to keep information from their child.
- The psychological needs of the adolescent are specific and complex. They may have difficulties dealing with the anxiety and uncertainty of the illness despite awareness of the prognosis.
- They will have concerns about their parents and siblings.
- They will have a desire for independence and a need for opportunities to do things that other young people do.
- School, college and employment will be of importance.
- Adult services are often inexperienced in dealing with the challenge of caring for adolescents.

(DOHC 2010)

In 2005 "A Palliative Care Needs Assessment for Children" was published and the following issues around the care for adolescents were raised:

- The need for:

    - Improved transition from paediatric to adult services
    - Improved facilities for adolescents when in hospital
    - More education for carers dealing with adolescents
- Opportunities for privacy
- Interaction and communication with trusted adults other than parents
- Involvement in collaborative decision-making with healthcare professionals and parents

(DOHC 2010)

1.14.5 What competencies are required when working with children with life-limiting conditions?

Healthcare professionals require the ability to:

- Assess the needs of the child and their family
- Respect and listen to the parents' knowledge, skills and choices
- Communicate with children and adolescents
- Deal with the specific problems of childhood illnesses and their management
- Work effectively as a member of an interdisciplinary team
- Provide support and care for the whole family
- Advocate on behalf of families to secure services from other agencies
- Work with families from various cultural and ethnic backgrounds, family structures and beliefs and acknowledge the influences these can have on the care of the child
- Maximise the developmental potential and quality of life of the child
- Proactively plan for potential problems in the near future
- They require the knowledge of the available facilities and how they can be accessed by families
- They need the appropriate skills to assess and manage:
    - Psychosocial issues of the children and their families
    - Symptoms during illness and at the end of life
    - The spiritual requirements of the children and their families

(DOHC 2010)

1.14.6 Palliative Care Services for Children with a Life Limiting Disease

- In Ireland, it is estimated that almost 1,400 children are living with life-limiting illness, and over 350 die each year (Irish Hospice Foundation 2013).
- For a service to provide optimal palliative care to a child with a life-limiting disease, they must comprehensively see to all the needs of the child.
- The Association for Children with Life-threatening or Terminal Conditions and their Families (ACT) in the UK have produced internationally recognised guidelines describing the level of palliative care service required to optimally cater for the needs of children with a life limiting disease The International expectation is that each country should provide palliative care services to meet these needs (DOHC 2010).
- Full details of the ACT Charter (2008) are available at http://www.dohc.ie/publications/pdf/palliative_care_en.pdf?direct=1

1.14.7 Palliative Care Services in Ireland

In Ireland, palliative care is provided to children with life-limiting conditions within the acute and community services. It is often delivered through and integrated with the existing child health services including the disability services. The *Palliative Care Needs Assessment for Children (2005)* comprehensively assessed the services and resources available to children with life-limiting conditions and their families. It was starkly evident that there was a lack of state services that specialise in PPC both in the hospital/hospice and the community setting. The report also found that the palliative care services for children in the community have evolved and developed differently over the last number of years which has resulted in some services being delivered directly by the HSE and others by voluntary agencies. Furthermore, there has been (and still is) a reliance on the voluntary sector to fill gaps in the palliative care services provided for children. While these organisations are very willing and critical to children's palliative care, they may lack the experience and competencies to provide the level of palliative care expected, as described by the ACT charter.

The DOHC (2010) document 'Palliative care for children with life limiting conditions in Ireland - A National Policy' provides full details on the palliative care services that exist in Ireland, available on http://www.dohc.ie/publications/pdf/palliative_care_en.pdf?direct=1 These services are:

1. Hospitals

There are currently 22 paediatric units attached to acute hospitals in Ireland, all of which provide some care to children with life-limiting diseases (DOHC 2010). These units have specialist workers who aim to meet the needs (other than health) of the children, including play needs and educational needs.

While efforts are made to make these acute hospital environments palliative friendly for both child and family, these paediatric units do not always provide the ideal environment for the optimal delivery of this service. In 2007, the 'Hospice Friendly Hospitals' programme was launched. This project aims to integrate hospice principles into hospital practice. The first and currently the only hospital to undertake this programme is Our Lady's Children's Hospital Crumlin which has the only specialist PPC team in Ireland. Further information can be found through the following link:
http://www.iapc.ie/iapc-directory-details.php?id=73.

2. Community and Home Care

The *Palliative Care Needs Assessment for Children (2005)* found that the preferred location of caring for a child with a life-limiting condition is the family home with parents receiving adequate professional support. Home was also the preferred location as the eventual place of death for a child. Despite this, due to several factors listed below, the majority of terminally ill children (except for children with cancer) continue to die in hospitals:

- Funding
- Reduced coordination of care in the home
- Lack of education and support to parents and carers
- Lack of formal training in palliative care for health professionals
- Complex and unpredictable diseases
- Access to aids and equipment and emergency resources

Currently the provision of palliative care services for children in the community is reflective of the child's diagnosis and care options available rather than the needs of children with life-limiting conditions. This care is being provided by healthcare professionals across a number of disciplines that are employed in the statutory or voluntary sectors. Full details of these are available on http://www.dohc.ie/publications/pdf/palliative_care_en.pdf?direct=1 and http://www.cancer.ie/publications/children-cancer

3. Hospices

At this time there is no National Children's Hospice in Ireland. The only children specific hospice in Ireland at present is 'LauraLynn House' based in the Children's Sunshine Home, Dublin. This hospice provides a range of services to children (0-18yrs) with life limiting diseases and their families. Information can be found through the link below:
http://www.lauralynn.ie/about.html

The Irish Hospice Foundation launched the 'Hospice Home Care Programme' based on the findings of the *Palliative Care Needs Assessment for Children (2005),* with the overall aim of providing a coordinated home focused palliative care service to all children with life limiting conditions in Ireland, and their families (Irish Hospice Foundation 2013).
Further information can be found through the following link:
http://hospicefoundation.ie/what-we-do/childrens-palliative-care/childrens-hospice-homecare-project/

4. Respite

Respite is defined as "The provision by appropriately trained individual(s) of care for children with life-limiting conditions, for a specified period of time, thus providing temporary relief to the usual care-giver".

(DOHC 2010)

Currently in Ireland, the availability of appropriate, child-friendly respite services for children with life-limiting conditions is limited. The few services that do exist are a combination of the public and voluntary sectors.
There are three main types of respite care:

- In the home
- Out of home
- Specialised

There are also three categories of respite which depends on the severity of the child's condition at any given time:

- Category 1: The provision of care by a trained/accredited healthcare assistant or a family member/friend.

- Category 2: The provision of care by appropriately trained healthcare staff with the requisite skills to care for a child with a life-limiting condition.
- Category 3: The provision of an extended range of respite services by an appropriately qualified and experienced registered nurse with specific expertise/qualifications in palliative care and paediatrics.

In-the-Home Respite:

These services provide temporary (average 3hrs) relief to families during the day.
The level of respite care provided in the home is divided into three categories:

The following are examples of in-the-home respite services:

- Irish Hospice Foundation: http://hospicefoundation.ie/what-we-do/childrens-palliative-care/
- The Carers Association: http://www.carersireland.com/services_homerespite.php
- LINK Service (Enable Ireland): http://www.enableireland.ie/content/respite-services
- The Jack & Jill Foundation: http://www.jackandjill.ie/

Out-of-Home Respite:

Here, families or just the child with a condition can take respite breaks throughout the year. Age appropriate activities which promote inclusion and independence are arranged prior to the child's arrival. Again, this respite service is broken into three categories:

Examples of out-of-home respite services:

- Silverpines Community House based in Bray (Enable Ireland), for children over the age of 7: http://www.enableireland.ie/content/respite-services
- Cheshire Ireland: http://www.cheshire.ie/services_ireland.php
- The National Home-sharing and Short-breaks Network (NHSN): http://www.nhsn.ie/
- Hospice services (see above)
- Make a Wish Foundation - This is a children's charity dedicated to granting the wishes of children aged between 3 and 17 years living with a life-threatening medical conditions: http://www.makeawish.ie/index.php?option=com_content&view=article&id=112&catid=9&Itemid=103

Specialised Respite:

Specialist respite care refers to a programme/setting of care that provides additional services. It may take place in the child's home or in a setting outside of the home such as a hospital, long term care facility or hospice. Specialist respite care provides the support required to meet the child's holistic care needs and enables children and families to access short break services. Specialist respite care will often address some aspects of symptom management. At present, specialist respite care is not available in Ireland. However, it is suggested in the report by the DOHC (2010) that there is a need for new or extended services to be developed, including statutory/voluntary partnerships.

5. Education and Training of health professionals

There are a number of training and educational services available to health professionals in palliative care which can be located through the following link: Database of Palliative Care & Bereavement Courses in Ireland

(Irish Hospice Foundation 2012)

However, these are not specific to PPC which as identified previously can be quite different with regard to management.

The *Palliative Care Needs Assessment for Children (2005)* identified a significant lack of training and education programmes for health care professionals in the area of PPC. Furthermore, there is no formal education of undergraduate health professional in the area of PPC. In order to overcome this problem, services to enhance the care given to children with palliative needs were and are being developed. They are outlined in DOHC (2010), available on: http://www.dohc.ie/publications/pdf/palliative_care_en.pdf?direct=1

6. Bereavement Support

The death of a child is often called the ultimate tragedy and is one of the most difficult deaths to grieve, as it goes against the natural order. There are three levels of support available to people who are bereaved:

General Support and Information (Level 1) – This applies to the vast majority of people who are going through the bereavement process. Information on things like knowing what the normal grieving pattern is and how to cope with the various different emotions can be very

important. The Irish Hospice Foundation as well as many other voluntary organisations and charities provide this information (links provided after next two sections). Forums, videos, booklets and more can all be found through these services.

Extra Support (Level 2) - This applies to a smaller percentage of people who need extra support with their grief. They may for example be struggling to cope because they don't have someone to talk to or may not want to confide in a family member. Most major **hospitals** and a large proportion of **hospices** provide acute bereavement support. Hospices will often provide further and on-going support and advice to those who require it in the days and months following a death. However, it is the voluntary bereavement support services, self-help groups, faith groups and community groups who provide the majority of the support at this level. Examples of such services include:

- Bethany Bereavement Support Group: http://www.bethany.ie/
- Barnardos: http://www.barnardos.ie/what-we-do/specialist-services/bereavement-counselling.html
- Rainbows: http://www.rainbowsireland.com/index.php?option=com_content&view=article&id=87&Itemid=205
- The Bereavement Counselling Service**:** http://www.bereavementireland.com/

Therapy Support (Level 3) - This is specialised support provided by professional (psychologists, psychotherapists, counsellors and doctors) and is appropriate for people who develop complications or become stuck in their grieving process (approximately 10-15%). This can often be the case with the death of a child. Although there is currently no professional bereavement counselling services in Ireland, the following services have professional workers who specialise in the area of grieving and bereavement:

- The Irish Association of Counselling and Psychotherapy (IACP)
- The Irish Association of Humanistic and Integrative Psychotherapy Ltd
- The Psychological Society of Ireland (PSI)

7. Information for Parents and Carers

The Children's Palliative Care Special Interest Group (SIG) was set up in 2009 as a branch of the Irish Association for Palliative Care (IAPC). It is representative of all the service providers of children's palliative care in Ireland (IAPC 2013).

Further information can be found through the following link: http://www.iapc.ie/childrens-palliative-care.php

1.14.8 Working as a Health-Care Professional in PPC

Working in a paediatric palliative care (PPC) setting and caring for children with life-limiting conditions is highly stressful (Liben *et al* 2008). Health care professionals (HCPs) caring for PPC patients face both mental and emotional challenges (Michelson and Steinhorn 2007). There is little known about how HPCs cope with the challenges of working in PPC, but studies suggest that they are at risk of developing burnout and compassion fatigue (Liben *et al* 2008).

- Burnout reflects movement 'away from caring and toward apathy'. It is often reflected in both physical and psychological symptoms, affecting overall well-being of the HCP. It often leads to poor job satisfaction and feelings that their work is less valued by others.
- Symptoms include: mental and physical exhaustion, insomnia, poor concentration, depression and loss of appetite and lead to reduced job motivation.

(Barnes 2001; Redinbaugh *et al* 2001)

When working with dying patients, feelings of powerlessness and a sense of failure often occur. HCPs may experience grief, self-doubt, fear of becoming ill themselves, and may even reconsider working with this patient group to avoid these feelings (Michelson and Steinhorn 2007).

Common stressors experienced by HCPs in PPC include:

- Exposure to childhood suffering and death
- Communication difficulties with young patients and parents
- Team conflict
- Inadequacy of support systems for care providers
- High workloads and staff shortages aggravating the above stressors

(Liben *et al* 2008)

1.14.9 Health-Care professionals and Grief

A number of work situations can create grief in the life of a HCP. Grief is the psychological distress associated with loss (Redinbaugh *et al* 2001).

The HCPs grieving process has unique characteristics common to all clinicians regardless of the different work environments and cultures. The grieving process is triggered by the loss the clinician feels when their young patient dies. The different types of losses include:

- The loss of a personal bond they have developed with the child
- The non-realisation of their efforts to cure or control the disease
- Unresolved personal loss that surfaces with the death of the child

(Liben *et al* 2008)

Characteristic of the HCPs' grieving process:

- Fluctuations between experiencing grief by focusing on the loss and avoiding/repressing grief by moving away from it. This fluctuation is healthy and adaptive. It helps the healthcare worker attribute meaning to the death of the child, and also their contribution to the healthcare of the child
- If the health professional remains submerged by their grief or suppresses/denies it and does not have fluctuating feelings complications can occur (Liben *et al* 2008)

1.14.10 Coping Strategies and Support for Health-Care Professionals

Coping refers to those behaviours that either reduce/eliminate the source of stress or mitigate the emotions elicited by the stressor (Redinbaugh *et al* 2001).

Personal coping mechanisms vary from person to person when dealing with emotional and morally challenging situations. Thus, a diversity of outlets needs to be provided by institutions to accommodate individual needs (Michelson and Steinhorn 2007).

Coping strategies include:

- Engaging in self-care activities such as exercise, meditation, or journal writing
- Developing a personal philosophy of care
- Engaging in self-reflection and self-awareness

- Committing to taking care of one's self
- Development of supportive professional relationships at work that promote debriefing and enhance mutual support
- Participation in support groups
- Organisation of regular debriefing sessions where practitioners can discuss cases
- Talking with a professional counsellor (if one is provided in your workplace)

(Liben *et al* 2008; Michelson and Steinhorn 2007)

1.10.11 Physiotherapy Treatment in Palliative Care

Physiotherapy role in palliative care:
Physiotherapists play an integrated part in the Multidisciplinary palliative care team from improving quality of life to symptom control in patients with life limiting conditions (Morrison *et al* 2005).

- The physiotherapist's main goal is to change the patient's behaviour in terms of encouraging and enabling independence, providing relief from distressful symptoms and offering support (Toot 1984).
- Physiotherapists in palliative care are involved in four levels: prevention, acute and post-acute care, community and institutional based rehabilitation and symptom control (Laakso 2006).

Symptom control issues:
The most common symptom control issues and functional disabilities in palliative care include pain, deconditioning, cough, weakness and breathlessness (Kumar *et al* 1996). In order to prevent and manage these symptoms the physiotherapist should aim to: Maintain optimum circulatory and respiratory function, prevent muscle atrophy and shortening, influence pain control, prevent joint contractures, optimize function and independence, and then education and participation of the family and/or carer (Kumar and Jim 2010).

Physiotherapy intervention:

The most common physiotherapy interventions carried out in the palliative care setting include: Therapeutic exercise, electrical modalities, thermal modalities, mechanical modalities, additional physical agents, miscellaneous modalities, manual physical therapy (myofascial, articular and neural) and assistive devices.

For more detailed information see: http://www.jpalliativecare.com/article.asp?issn=0973-1075;year=2010;volume=16;issue=3;spage=138;epage=146;aulast=Kumar

Quality of life (QOL)

Maintaining and improving the QOL of young people is the main goal in PPC. It is a misconception that palliative care is only and can only be delivered at the end of life stage of a patient, when in fact it can be delivered at any point in the disease trajectory of the child with a life shortening neuromuscular disease, parallel with the active treatment they are receiving.

Assessing the QOL of a child can be challenging as the gold standard for assessment of HRQOL is self-report. Often there can be inconsistencies in the level of agreement between the carers/parents and child. Instruments used to assess QOL should be age specific and based on self-report.

Ethical use of Interventions:

NIV can be used for symptom management in respiratory failure where the goal is not to extend life. Relieving the symptoms of dyspnoea and the consequences of carbon dioxide retention can improve QOL. The weighing of 'benefits' and 'burden' is involved in the decisions regarding long term ventilation for each individual. By their very nature they are value laden and cannot be made unilaterally.

A sound 'transparent' and ethical approach would be to recognise that the healthcare providers' own attitudes and beliefs can be offered in conjunction with the proposed treatment plan ideas and options. This allows deliverance of factual information in conjunction with the healthcare providers' own values and experiences in a specific context.

Recommendation for therapist:

- In assisting patients, parents and carers in making informed choices that are also consistent with their own preferences and values, physicians are required to engage

patients and their parents/carers in a process of equal participation in decision-making including full disclosure of all information in a timely and sensitive fashion.

- An open discussion with all the multidisciplinary team regarding the duration and type of specific interventions can encourage shared decision-making and transparency.

Psychological and Mental Health Issues:

There is a definite need for more mental health support available for children and young people that have a neuromuscular disease and their family/carers.

There have been reports of increased maladaptive coping strategies, mental health issues and behavioural problems within this population and their parents/carers.

Recommendation for physiotherapist:

- The benefits and burdens of all possible interventions must be looked at and considered with relation to the impact and mental health of the patients and their families.
- The therapist should always be aware of and acknowledge that patients and/or their families may be under stress, have mental health issues or require further support from organisations.

Symptom Management and End of Life Care:

Children and young people with neuromuscular disease (NMD) are significantly burdened through life with physical symptoms. Prolonged survival comes with an increase in morbidity of respiratory and non-respiratory issues. There is a limited amount of evidence available regarding end of care life in children with NMD. Often terminal respiratory failure is the mode of death.

General Symptom Management:

In a national survey of hospice provision in people with NMD, it was found that boys with DMD most often require some form of symptom management and medical intervention during a routine respite stay (Fraser *et al* 2011).

Dyspnoea:

- Non-drug management of dyspnoea includes secretion management, an open window or use of fan, massage or visualisation for anxiety, and appropriate positioning for the child. Nearing the end of life, families may request to remove NIV equipment from the child's face. For this to happen there must be sensitive discussion, careful timing and prompt symptom management.
- Drug management involves careful use of opioids and psychotropic medication. Low dose opioids are used to manage distressing breathlessness even when active treatment is pursued, Benzodiazepines are used for agitation.
- With inter-current infection or aspiration, breathlessness is a distressing symptom that can arise. These often occurs in conjunction with progressive neuromuscular deterioration, cardiac failure and in the final stages of the disease. Symptoms of respiratory distress can be effectively palliated with NIV, and effective secretion clearance and feeding management.

Pain:

A significant link has been reported between increased pain and fatigue and decreased levels of general health and social function. Chronic pain is common in neuromuscular conditions and poor pain management is negatively correlated with QOL and can impact on various aspects of care, in particular respiratory care.

Table 1.14.2 Signs and Symptoms

Symptoms of respiratory failure	Symptoms of nocturnal hypoventilation	Symptoms of Bulbar Dysfunction	Signs of respiratory muscle weakness
• Weight loss • Orthopnea (Dyspnoea when supine/prone) • Lethargy • Recurrent chest infections	• Fatigue • Reduced concentration • Un-refreshing sleep • Frequent nocturnal wakening • Excessive daytime sleepiness • Early morning headache	• Weak cough • Recurrent chest infections • Nasal regurgitation • Choking or coughing episodes at mealtimes	• Increased rate of respiration • Reduced chest expansion • Cyanosis • Weak sniff or cough • Abdominal paradox • Recruitment of accessory muscles at rest • Pallioedema (severe hypoventilation)

Advance Care Planning:

- It is essential for the MDT to involve the patient and their family/carers in a process of mutual decision making in order to make informed decisions.
- Discussions with different healthcare professionals, support groups and peers/ parents of children with the condition can help to consolidate the shared decision-making process. To support this process, choice promotion through continued assessment of need, high quality provision of information and open discussion at key points during disease trajectory is needed.
- Advance care planning is a 'voluntary process of discussion and review to help anticipate how an individual's condition may affect them in the future'. An advanced care plan is not a legal document or a 'do not resuscitate' order. It changes in parallel with the progression of the disease.
- In paediatric practice, it incorporates wishes for emergency and ensures written information regarding future clinical management documented. The main aim is to give the patient and their family an opportunity to plan end of care life care and have a dignified and symptom free death.

(Weiner *et al* 2008)

Recommendation for Physiotherapist:

- The MDT should review advance care plans on a regular basis.

1.15 Key References

- Department of Health and Children (DOHC) (2010) *Palliative Care for Children with Life-Limiting Conditions in Ireland – A National Policy* [online], available: http://www.dohc.ie/publications/pdf/palliative_care_en.pdf?direct=1 [accessed 30 Jan 2013].
- National Council for Special Education (NCSE) (2011) *Children with Special Educational Needs – Information Booklet for Parents* [online], available: http://www.ncse.ie/for_parents/Information_Pamphlets.asp [accessed 17 Feb 2013].
- Citizens Information (2013) [online], available: http://www.citizensinformation.ie/en/ [accessed 26 Mar 2013].

2.Down syndrome

(DS)

2.1. Background and Diagnosis

2.1.1. Pathophysiology:

95% of cases are caused by trisomy of chromosome 21 (Chen *et al* 2012). An extra chromosome 21 is found in all cells as a result of non-disjunction during meiosis in one of the parents. They have 47 chromosomes instead of 46.

Translocation and mosaicism account for the remaining 5% (Van Cleve and Cohen 2006).

2.1.2. Clinical Features:

- Decreased muscle tone
- Hearing problems
- Gastrointestinal
    - oesophageal atresia
    - duodenal atresia
- Hip problems- risk of dislocation
- Sleep aponea (mouth, throat, and airway are narrowed)
- Underactive thyroid

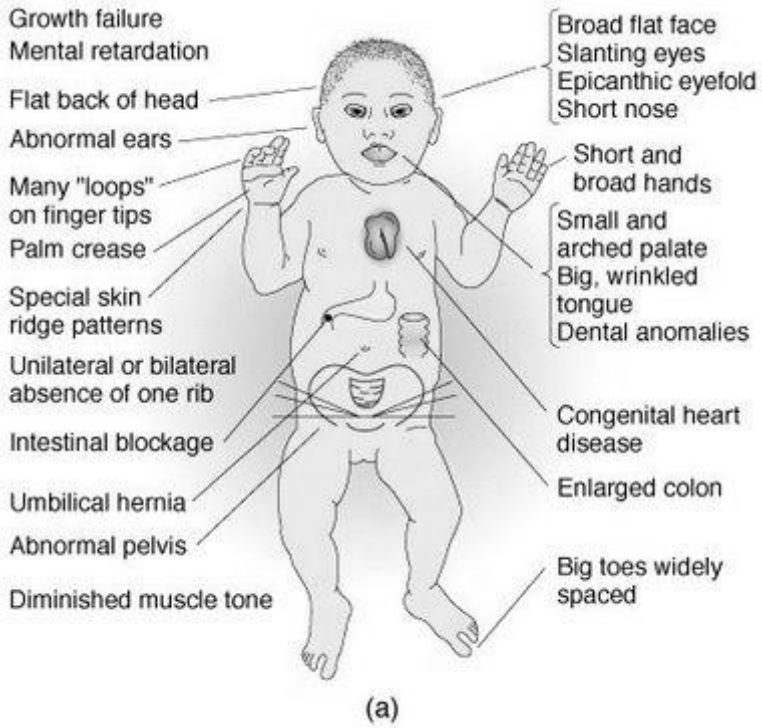


Figure 2.1.1 Clinical features of DS

2.1.3. Etiology:

DS is usually caused by abnormal meiosis on the maternal side.

Maternal age is a risk factor (Chen et al 2012) for DS. Prevalence ranges from 0.7/1000 births for mothers aged 20–24 years to 55/1000 births for those aged 45–49 years.

2.1.4. Epidemiology:

DS is the most common congenital cause of developmental disability in Ireland with a prevalence of 1 in 546 live births (DSMIG 2009).

Internationally it is diagnosed in 1 per 650–1000 live births (Bittles 2006).

2.1.5. Diagnosis:

Pre-term:

- Diagnostic
  - Amniocentesis
  - Chorionic Villi sampling
- Screening
  - Ultrasound
  - Maternal Serum screening testing

At Birth:

- Initial diagnosis at birth based on physical features.
- A blood test can be done to check for the extra chromosome and confirm the diagnosis.

2.1.6. Prognosis:

Live into adulthood with some limitations but defects may reduce life expectancy.

Life expectancy has increased in recent years due to improved healthcare.

2.2. Physical Management

2.2.1. Musculoskeletal

Developmental delay

Table 2.2.1. Gross motor developmental milestones for DS (DSMIG 2011)

	Down Syndrome		Normal development	
Activity	Average age (months)	Age range (months)	Average age (months)	Age range (months)
Holds head steady when sitting	5	3-5	3	1-4
Rolls over	8	4-12	5	2-10
Sits alone	9	6-16	7	5-9
Stands alone	18	12-38	11	9-16
Walks alone	23	13-48	12	9-17

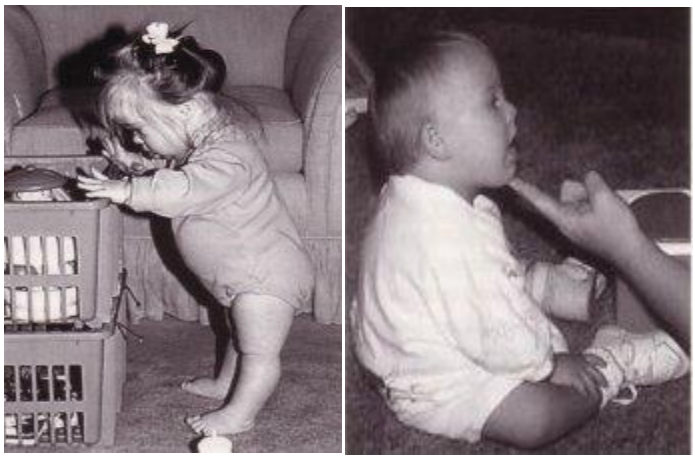


Fig. 2.2.1. Typical presentation of standing and sitting

- Physiotherapy Assessment of Motor Skills
    - Bayley Scales of Infant Development II Motor Scale
    - Peabody Developmental Motor Scales-2
    - Both scales have been shown to be valid, reliable, standardised and norm referenced (Provost et al 2004).
- Approach/Intervention:
    - Neurodevelopmental Therapy (NDT):

Fig. 2.2.2. Neurodevelopmental therapy

It is a "non-invasive programme of movements which aims to promote development of the nervous system and the inhibition of primitive reflexes" (The Irish Association of Neuro Developmental Therapists 2012). The child's strengths and impairments are identified and treated with regard to functional abilities and limitations (NDTA 2012). A systematic review concluded that NDT did not show a measurable positive effect early in life (Blauw-Hospers and Hadders-Algra 2005). However this review contained limited studies on NDT in infants with DS and only looked at studies with participants from 0-18 months. Overall there appears to be limited studies looking at the effects of NDT in DS for early intervention (age 0-5).

    - Treadmill Training:

The use of balance supported treadmill training to help with the acquisition of independent gait is based on the use of the stepping reflex; it has been shown that it is possible to elicit a pattern of behaviour similar to walking (Ulrich et al 2001). Ulrich et al (2001) used a home based treadmill training intervention and concluded that the use of a motorised treadmill with

balance support in the deliberate practice of alternating steps prior to independent gait can reduce the delay in acquiring this skill in infants with DS (Ulrich et al 2001). A further study investigated the effects of intensity of treadmill training and found that the higher intensity training group acquired the skill of independent gait earlier than their lower intensity trained counterparts (Ulrich et al 2008). Many of the differences in motor milestones however weren't statistically significant. Both high and low intensity treadmill training groups walked independently earlier than those receiving physiotherapy only.

Muscle Strength

- Muscle strength in both the upper and lower limbs is up to 50% weaker in individuals with DS in comparison to their peers (Sheilds and Taylor 2010).
- Muscle weakness is also associated with an increase in incidence of osteoporosis and reduced cardiovascular fitness (Angelpoloulou *et al* 2000).
- It reduces the ability to maintain balance (Carmeli *et al* 2002).
- Participation in work activity tends to be physically based therefore muscle weakness can negatively impact on their vocational performance (Sheilds and Dodd 2004).
- Management:
    - Exercise programs focusing on increasing muscle strength can result in significant increases in strength and can impact positively on participation in functional activities and physical skills in the work place (Carmeli *et al* 2002).
    - Participating in strength training may be more harmful than beneficial. They have associated muscular hypotonia and joint ligamentous laxity. Participating in a strengthening program that involves repetition of high resistance exercises may increase the risk of attaining a musculoskeletal soft tissue injury, joint subluxation or dislocation (Sheilds and Dodd 2004).
    - By introducing an exercise program to adolescents with DS, it can help establish good exercise habits which are positive predictors of healthy, regular activity in adults with DS (Telema *et al* 2005).
    - There is limited research to date that looks specifically at the effects of progressive resistance training in individuals with DS.

Table 2.2.2. Programmes delivered in studies

	Shield and Taylor 2010	Gupta et al 2010
Frequency	10 weeks • twice a week	6 weeks • 3 times a week
Intensity	• 3 sets of 12 repetitions or else • 3 sets to fatigue Resistance was increased once the sets could be done at ease.	• Strength exercises were carried out using a sand bag measured at 50 % of the individuals 1 rep max • 2 set of 10 repetitions carried out at each muscle group • Resistance was increased by 0.5kg when the participant could carry out the sets at without undue stress.
Time	2 min break between each set	• Necessary time to complete sets.
Type	<u>Progressive Resistance Training</u> 6 exercises using weights machines. 3 upper limb: • lat pull down, • seated chest press, • seated row. 3 lower limb: • seated leg press, • knee extension • calf raises	<u>Progressive Resistance Training</u> Exercises for the lower limb: • Hip - flexors, abductor, extensors, • Knee - extensors and flexors, ankle plantar flexors
Results	An average increase of 42% in muscle strength of the major antigravity lower limb muscles (hip extensors and quadriceps)	Significant increase in muscle strength.

Balance

- Previous research has proposed that motor dysfunction in children with DS was due to joint ligament laxity and muscle hypotonia. However, more recent research has argued that deficits in balance, control mechanisms and higher level postural control mechanisms may be the cause (Wang and Ju 2002).
- Poor balance and postural control leads to an increased difficulty in independence carrying out of functional ADL's (Carmeli *et al* 2002).
- Shumway-Cook and Woollacott (1985) report that children with DS consistently score lower on gross motor skills than their peers without DS, especially in static and dynamic balance. Similarly, Rahman and Shaheen (2010), report that children with

DS have lower scores than their peers on primarily balance and agility tasks and also running speed, strength and visual motor control.

Management:

- Balance training
  - Shumway-Cook and Wollacott (1985) report that in order to improve stability and balance the intervention should focus on improving automatic responses to postural control rather than conscious responses.
  - Gupta et al (2010) demonstrated that this approach is appropriate

Table 2.2.3. Programme delivered by Gupta et al (2010)

	Balance
Frequency	6 weeks 3 times a week
Intensity	• 10 repetitions of each exercise • increased by 5 when individual could complete with ease
Time	• Necessary time to complete sets.
Type	<u>Balance specific training</u> • Vertical jumps • Horizontal jumps, • Tandem stance, • One leg stance with eyes open, • Walking on a line, • Standing on a balance beam with eyes closed • Walking on a balance beam • Jumping on a trampoline.
Results	• Significant increase in scores on the balance subscale of BOMPT (10.5 →19.5) • No significant difference in 2 components – walking on a straight line and standing on a balance beam with eyes closed.

Atlantoaxial instability (AAI)

- 13% of those with DS have an increased atlanto-dens space but no associated symptoms. An additional 2% develop signs and symptoms of spinal cord compression (Roizen and Patterson 2003).

- For children with DS that have no symptoms associated with AAI, there is no evidence that participation in sport increases the risk of spinal cord injury (DSMIG 2009).

2.2.2. Cardiovascular

Heart defects (Van Cleve et al 2006)

- Childhood
    - Cardiac defects affect approximately 50% of children with DS.
    - Atrio-ventricular defects are the most common condition.
    - Some also suffer from ventricular septal defect, patent ductus arteriosus, atrial septal defect.
- Medical Management:
    - Screened at birth for cardiac defects and then a cardiac evaluation and echocardiogram are recommended if a murmur or click is found on physical examination.

Aerobic fitness

- Individuals with DS have lower levels of cardiovascular fitness then their peers (Fernhall et al 1996) due to a sedentary lifestyle and physical impairments.
- They have an increased risk developing osteoporosis, obesity, cardiovascular disease and type 2 diabetes (Fernhall et al 1996).
- It can negatively affect their participation in social, occupational and recreational activities (Dodd and Sheilds 2005).
- Management:
    - There is limited amount of research carried out on the benefits of cardiovascular exercise for individuals with DS, however, in other populations it has been shown to improve cardiovascular fitness, reduce mortality, increase exercise endurance and work capacity, increase muscle strength, assist with weight loss and control, and improve psychological well-being and quality of life (Van den Ende et al 2000, Rees et al 2004, Jolliffe et al 2001).
    - In a systematic review carried out by Dodd and Shields (2005) which included 4 studies, they concluded that exercise programs aimed at improving cardiovascular fitness can be very beneficial and effective for individuals with DS. Overall they found significant improvements reported by increases in the

maximum workload achieved, time to exhaustion, Vo_2 peak and peak minute ventilation.

- Physiotherapy intervention
    - A cardiovascular exercise program for individuals with DS as recommended by The American College of Sports Medicine (ACSM) guidelines.

Table 2.2.4. Aerobic exercise recommendations by ACSM

Frequency	3-7 times a week
Intensity	Aerobic activity carried out at 55-90% of peak heart rate or 40-85% of max oxygen uptake reserve **
Time	20 – 60 minutes
Type	Any aerobic exercise – Jogging, Walking, Rowing , Cycling

** It is still uncertain whether these are safe and beneficial for the individual with DS. Some authors suggest that physiological impairments that are associated with DS (such as lower Vo_2 peak levels and peak heart rate) may reduce their ability to improve their cardiovascular fitness levels (Fernhall and Pitetti 2001). Others however suggest that these programs can have significant effects on improving the physiological issues and hence improve their cardiovascular fitness (Rimmer et al 2004). Dodd and Sheild (2005) recommended the following training protocol based on the findings from their systematic review on cardiovascular exercise programs for people with DS.

Table 2.2.5. Recommendations made by Dodd and Sheild (2005).

Frequency	12-16 weeks • 3 times a week
Intensity	• 50-75% of Vo_2 peak
Time	• 30 minutes
Type	Jogging, cycling and rowing are the most common activities done and yield positive changes in fitness.

2.2.3. Gastro-intestinal

Congenital anomalies (Pueschel 1990)

- Observed in 12% of infants with DS
- They include:
    - Tracheoesophageal fistula
    - Pyloric stenosis
    - Duodenal atresia
    - Annular pancreas
    - Aganglionic megacolon
    - Imperforate anus
- Most require surgery so that nutrients and fluids can be absorbed

2.2.4. Vision

Cataracts

- Congenital
    - Are present in 3% of neonates
    - Examination at birth and removal if present
    - Correction by glasses

Management

- Regular examination by paediatric ophthalmologist
- In adulthood examination by ophthalmologist every 2 years (Smith 2001)

2.2.5. Other

Communication

- Communication is a common problem in DS due to intellectual disability and speech and language deficits. The main speech and language problems involve language production, syntax and poor speech intelligibility (Roberts et al 2007).
- Factors leading to poor communication:
    - Hearing loss:

        - This occurs in about two-thirds of children with DS. It can be caused by sensorineural or conductive hearing losses or both.
        - Otitis media, an infection of the middle ear commonly seen in children with DS, often causes fluid build-up in the middle ear. This can cause mild to moderate fluctuating hearing loss (Roberts et al 2007).
    - Structural and functional differences in oral structures:
        - These differences include a small oral cavity, a high arched, narrow palate, irregular dentition, and a large tongue that protrudes forwards.
        - Abnormal facial musculature including missing, additional, or poorly differentiated muscles and nerve innervation differences.
        - These impact on speech intelligibility by contributing to reduced speed, limited range of motion, and difficulty with coordination (Roberts et al 2007).
- How Children with DS and Speech Difficulties Communicate (Roberts et al 2007):
    - In typically developing children the pre-linguistic stage (the period before language is used), occurs from 12 to 18 months of age.
    - Communication at this stage is through gestures, vocalisations, facial expressions, and other movements.
    - This pre-linguistic stage can last for several years in children with DS.
    - Gestures are often used in the early stages of communicative development in children with DS.
- Interventions:
    - Augmentative and Alternative Communication (AAC)
        - AAC is an intervention approach that uses “manual signs, communication boards with symbols, and computerized devices that speak and incorporate the child’s full communication abilities” (Romski and Sevcik 2005).
        - AAC interventions are a tools that aids the development of early language skills.

        - Regardless of whether the child eventually talks or not, AAC helps to set the stage for later vocabulary development and combinatorial language skills (Romski and Sevcik 2005).
        - AAC supports speech development and best evidence indicates that it does not have a negative impact on speech production (Millar *et al* 2006).
        - It is thought that the use of AAC may allow individuals with developmental disabilities, such as DS, to bypass the cognitive and motor demands of speech production and instead focus on building communication and language skills (Millar *et al* 2006).
    - Lámh
        - Sign language is a commonly used form of communication for children with DS.
        - It is based on Irish Sign Language (ISL) and consists of 500 signs.
        - Speech is always used with Lámh signs and only the key words of a sentence are signed (Lámh 2013).
        - Lámh signs are used to support communication and are useful for many reasons, including the following (Lámh 2013):
            1. Eye contact and attention to movement are encouraged which are important for speech development.
            2. They can hear what is being communicated
            3. The Lámh user is more easily understood if they have limited or unclear speech.

Obesity

- Individuals with DS have a higher incidence of obesity than other individuals due to decreased activity levels, a lower resting metabolic rate and an unbalanced diet.
- While infants with DS tend to be light for their height initially, they soon progress to being proportional, then become overweight, and are more likely than not to be obese by the age of 3-4 years (Roizen and Patterson 2003).
- Those who are overweight in childhood tend to be overweight as adolescents and adults.

- 38% of adults with ID including those with DS were overweight and 26% were obese (De Winter et al 2012, Melville et al 2008)
- Being overweight and obese predisposes these individuals to hypertension, cardiac disorders, Type II diabetes and decreased life expectancy (Van Cleve et al 2006)
- The DSIMIG (2009) guidelines recommend that height and weight is recorded and charted using DS specific charts for the first 24 months of life.
- From 24 months of age, a lifelong regimen to monitor growth and prevent obesity should begin, including behavioural interventions, food selections, and physical and social activities (Roizen and Patterson 2003).

Hypothyroidism

- Prevalence of 16-20% (Van Cleve et al 2006)
- Can appear in childhood or develop in adulthood
- Management
    - Testing of TSH should be carried out if symptoms appear in childhood and routinely once a year in adults (Smith 2001)
    - Thyroid hormone treatment

Type II diabetes

- Prevalence rate of 1.4-10.6% (Van Cleve et al 2006)

2.2.6. Other issues and interventions that a physiotherapist should consider

Pre-exercise considerations (Sanyer 2006, Murphy and Carbone 2008)

- Determine health status prior to participation
- Consider the level of competition they wish to compete at
- What Sport do they want to play and what position do they play?
- What protective/adaptive equipment will they need to aid participation?
- Can the sport and rules be modified to aid inclusion?
- What ability has the child and the parent to understand the risks?

Combined training:

- There is a limited amount of research carried out on the effect of combining these interventions.
- Aerobic and Resistance:
  - Mendonca *et al* (2011) looked at the effects of combining aerobic training with resistance training in adults with and without DS.
  - After a 12 week intervention there was an improvement in physical fitness, walking economy, the ability to perform ADL's and an increase in work capacity.

Table 2.2.6. The exercise training program that Mendonca *et al* 2010 followed

	Resistance	Endurance
Frequency	12 weeks • 1 day a week	12 weeks • 1 days a week
Intensity	2 x circuit rotation of 9 exercises: • 1 set of 15 repetitions of abdominal curls • 8 dynamic exercises. • All exercises were prescribed at 12 RM for each individual participant (determined pre the intervention) • When the participant could perform the circuit with proper lifting technique and no breath hold, their load was increased by 10%.	• Aerobic exercise at a target heart rate of 65-85% Vo_2 peak
Time	• 30 seconds rest between each exercise	• 5 minutes warm up • 30 minutes main set • 5 minutes recovery
Type	• abdominal curls 8 dynamic exercises: • chest press • leg press • vertical traction • lower back • leg extension • triceps push down • biceps curl	• treadmill walking or running • 0% incline

- Rimmer *et al* (2004) incorporated progressive resistance training into an aerobic intervention and showed an increase in Vo_2 peak values. As there is a strong correlation between leg strength and Vo_2 peak in individuals with DS (Pitetti and Boneh 1995).

  - Strength and Balance:
    - Gupta *et al* (2011), combined strength and balance training in children with DS.
    - After a 6 week intervention of progressive resistance training for the lower limb and balance exercises, a significant improvement was found in both lower limb strength and overall balance.

Individual VS Group Exercise

- There are no studies that examine whether individual exercise programs or group exercise programs yield better result for individuals with DS specifically.
- Menear (2007) conducted a qualitative study (focus groups) and looked at parents perspectives of children with DS and their participation in physical activity.
  - Parents of primary school age identified a need for group activities
  - Group participation with siblings was highlighted as a motivating factor
  - Parents of adolescents identified a need for teenagers with DS to be involved in an individual sport that does not require that their opponents and team mates to be ability matched.
- Research in other populations:
  - No studies have found that one type of program is more beneficial than the other (Floyd and Moyer 2010, Tiffreau *et al* 2007)
  - Johnson (2009) - A Systematic review of children with a disability
    - Moderate evidence for group fitness interventions in developmental disability as well as DS.
    - Reported to improve CVS function, gait speed, functional mobility, self-perception, and satisfaction.
    - Children were motivated by their peers.
    - There are financially feasible approaches in a paediatric population

2.3 Psychological Management

2.3.1. Psychological wellbeing of family

Family reaction to diagnosis and coping

- Positives and negatives coexist in most situation
- Perceived positives and negatives are affected by the coping strategies and appraisal of the stressor by the individual

Table 2.3.1. Parent and sibling coping of a child with DS

	Parent	Siblings
Positives	• Enjoy their role • Cope well with increased demands • Develop mastery in new role • Develop greater self-esteem • Embrace new perspective on life • Maintain hope and defiance for future of child psychological growth • improved relationships learned patience and acceptance improved understanding of love and advocacy • Family functioning not different to norms • (Skotko et al 2011)	• They become empathetic, responsible, caring and independent (Hodapp et al 2007) • less externalizing and internalizing problems than other disabilities (Hodapp et al 2007) • no negative effect on behaviour or perception of competence (Hodapp et al 2007, Cuskelly et al 2006) • 11/23 would not change anything about their life (Graff et al 2012). Those that would change aspects mentioned were communication, behaviour and their own personality • No difference in academic or social performance (Cuskelly et al 2006)

Negatives	• Stress (Dabroska and Pisula 2010) • Due to: financial reasons, child needs ,uncertainty about prognosis, social ostracism, negative appraisal of child, • lack of family communication, depression • More marital conflict (Hodapp et al 2007)-divorce higher in early years (32% before age of 2) but less likely compared to other disability (6%) QoL of family decreases as severity of disability increases (Davis and Gavidia-Payne 2007)	• Higher risk of anxiety and depression • Less attention from parents (Skotko et al 2011) • Higher caregiving duties scores - care-giving increased with sibling age (Cuskelly et al 2006)
Feelings toward child (Skotko et al 2011)	Majority of parents: o Love o pride Minority of parents: o Embarrassment- increased with increased learning difficulties o Regret – associated with having other children with disabilities	Majority of siblings: o Love o pride Minority of siblings: o Sad o Feel sorry for child o Worried that they would be teased o Embarrassed
Advice to future parents (Skotko et al 2011)	• Joy outweighs struggle • Life goes on • Seek resources and support • Learn to teach and advocate for your child	• Joy outweighs struggle • Life goes on • Seek resources and support • Learn to teach and be patient • Don't worry about what others think (Graff et al 2012)

Coping strategies:

- Effective coping strategies (Davis and Gavidia-Payne 2007)
  - Social support - lowers stress and difficulties
  - Parental perception of control
  - Grieving (Barnett et al 2006)
    - Grief for expectations and hopes
    - Allows secure attachments to form
    - This is an on-going process
  - positive appraisal of situation
  - They cope better than families of other disabilities due to responsiveness of child and better support services

- Ineffective coping strategies:
  - remain preoccupied with negative reactions (Barnett et al 2006)
  - deny any negative feelings or disappointment
  - Not dealing with grief – 50% remain unresolved at 2years post Dx, time is not a sufficient healer (Barnett et al 2006)

2.3.2. Psychological wellbeing of adolescents with DS

Effect of the transition to secondary school

- Transition from primary school to secondary school for those with an intellectual disability can often have a greater impact on mental health (intellectualdisability.info 2013).
- Pupils with DS find adapting to change and adjusting to new surroundings difficult (DSA 2013).
- Student's perceptions of the quality of their school life decreased during this transition (Maras and Aveling 2006).
- No difference in the prevalence of psychiatric disorders was found between adolescents with an intellectual disability attending mainstream schools and those attending special schools (Emerson 2003).

- If a student is having difficulties with the transition it can manifest itself in several ways including (Maras and Aveling 2006):
    - sudden outbursts of inappropriate behaviour
    - chronic illness
    - detachment from the new environment
    - negative effects on academic performance, self-perceptions and perceptions of school

Other issues:

- Mental health and psychiatric illness
    - Mental health problems are experienced by four out of ten adolescents with an intellectual disability (intellectualdisability.info 2013).
    - They have a significantly increased risk of psychiatric disorders (Emerson 2003). This is based on a limited number of prevalence studies.

- Friendship and social inclusion
    - Adolescents with just a mild intellectual disability experiencing stigma for being "different" often use strategies such as denial, unrealistic self-appraisal and minimisation of the disability, and overcompensation.
    - These behaviours can lead to social isolation if they reject those similar to them and are rejected by their non-disabled peers (Cunningham and Glenn 2004).
    - Many adolescents had difficulties forming friendships that continue outside of school hours (D'Haem 2008)
    - D'Haem (2008) reports cases where adolescents with DS become depressed due to social exclusion outside of school hours

- Useful resource for parents to help with the transition process

- http://www.down-syndrome.org/practice/2016/
  http://www.downsyndrome.ie/index.php/general-educational-information/transition-to-post-primary-school

2.4. Promoting independence and participation in the community

2.4.1. Physical activity (PA) and leisure activity participation

Get Ireland Active physical activity guideline:

- It is recommended that children participate in at least 60 mins of moderate to vigorous physical activity per day. This should include bone-strengthening, muscle-strengthening and flexibility.
- There are no guidelines for children with disability in Ireland.

Participation pattern of individuals with DS and other disabilities
Down syndrome

Children

- Limited studies available in this population
- 58% of children with DS do not meet the above recommendations and none participated in vigorous activity (Shields et al 2009).
- The main source of PA reported by parents is PE and unstructured play at home (Menear 2007).
- They do not initiate physical activity/play themselves and rely on siblings and peers to include them (Menear 2007).

- One study showed that they choose to participate in activities that are consistent with the strengths of their phenotype i.e. they participate in visual-spatial activities but avoid physical and musical activities (Sellinger *et al* 2006)
- There was a gender difference in some activities with boys participating in more physical tasks while girls participated in more social activities but this was only reported in small study conducted in Taiwan (Wuang and Su 2011).

Adolescence

- As reported in the general population there was a decrease in activity with increasing age (Shields et al 2009)
- Adolescents participate mostly in social and skill-based activities and in the informal activities due to higher enjoyment and social engagement in these activities (Wuang and Zu 2012).
- However there are limited studies on adolescents with DS and PA.

Participation of youths with Intellectual Disability:

- A review of PA in children with ID including children with DS concluded that there was no clear pattern or parameters to participation due to contradictory results in the literature (Frey et al 2008).

Participation of youths with a disability: (Rimmer and Roland 2008)

- Youths with a disability have a 4.5 times higher rate of physical inactivity compared to non-disabled peers.
- Adolescents were twice as likely to report watching tv for more than 4 hours a day than their non-disabled peers.
- Disabled children spent more time in sedentary activity and less time engaged in team sports.

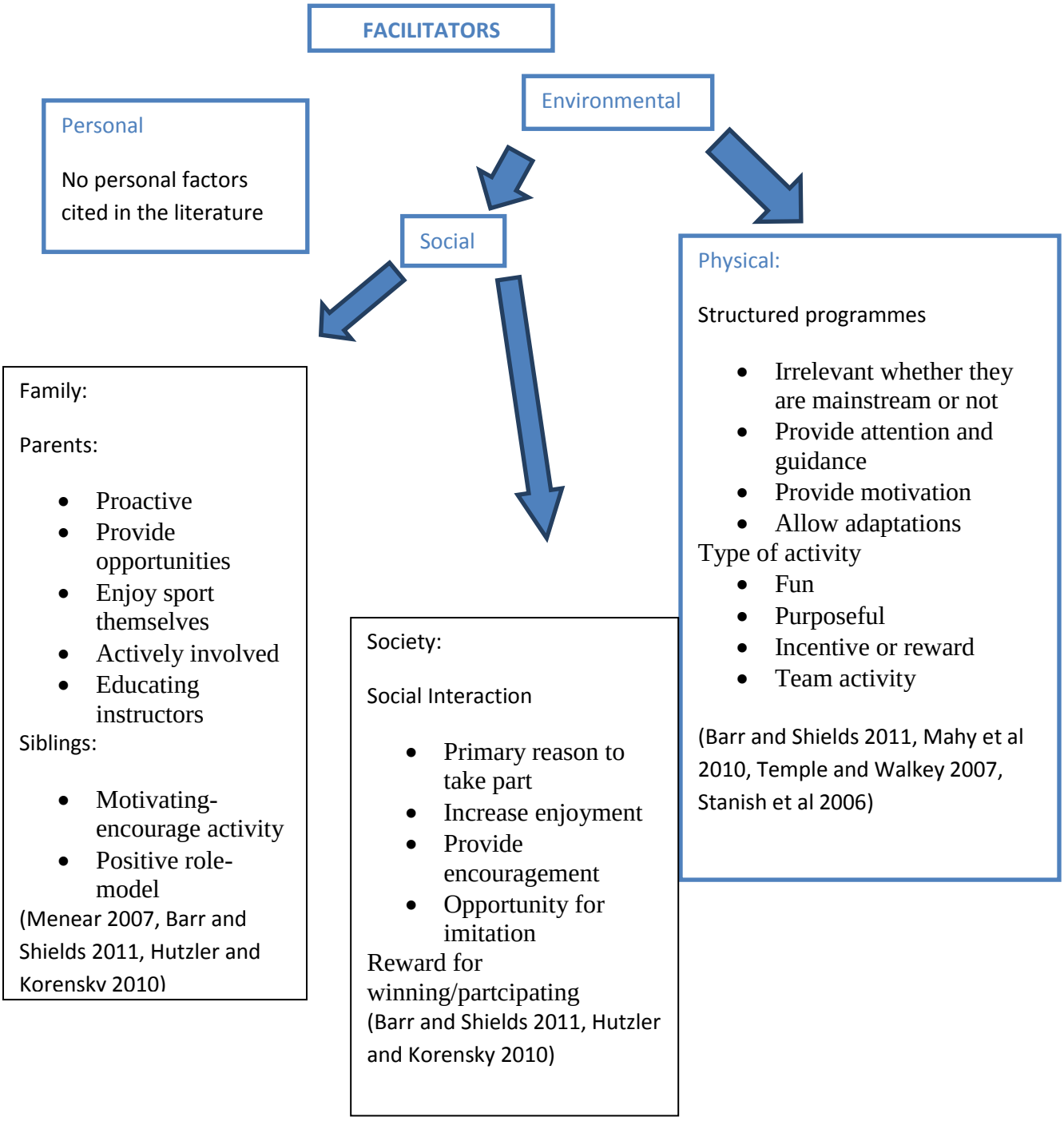
Importance of PA

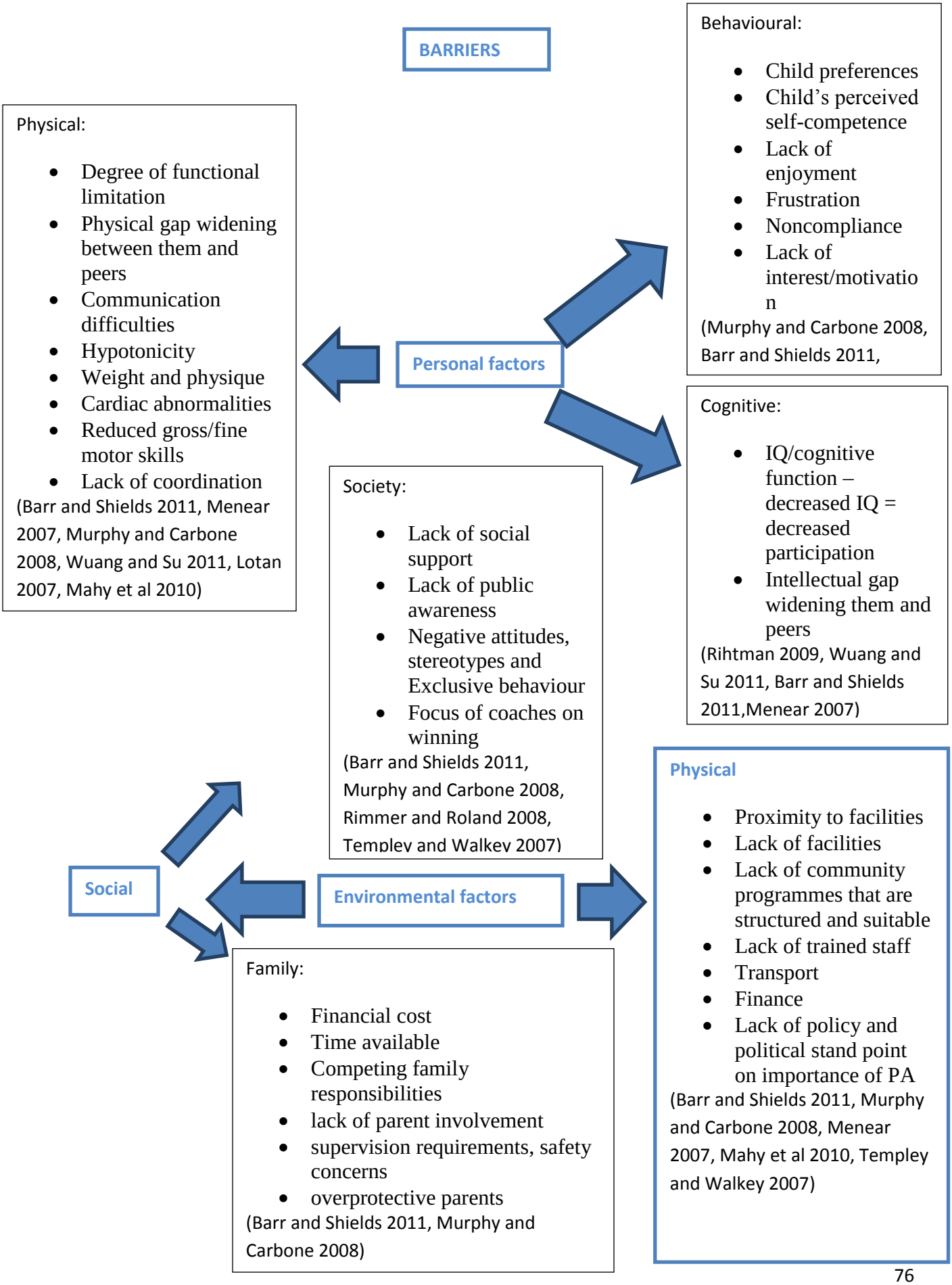
- For children with a disability (Murphy and Carbone 2008)

    - Physical inactivity predisposes children to obesity, type II diabetes and cardiovascular disease
    - Participation is the context that individuals form friendships, express creativity, achieve mental & physical health & develop meaning and purpose in life.
    - Reverse physical deconditioning
    - Increases exercise endurance and cardiovascular fitness
    - Reduces risk of obesity
    - Increases psychological well-being and social activity
    - Foster independence, coping, competitiveness and team work
- Children with DS
    - All the above reasons apply
    - Higher rate of obesity in this population
    - They have lower CVS fitness
    - They have reduced muscle strength
- The Healthcare system and country as a whole (Stanish et al 2006)
    - High healthcare cost associated with inactivity
        - $75 billion in US per year
        - No figures for Ireland
    - Indirect costs associated with inactivity such as immature death, lost wages and work limitations
        - Costs $38million in the US per year
        - Accounts for 76% of total lifetime costs for illness related to an ID diagnosis

Barriers and facilitators of participation in PA

According to the World Health Organisation (2001) and the International Classification of Functioning Framework participation is affected by personal factors, as well as social and physical environmental factors.


FACILITATORS
Personal
No personal factors cited in the literature
Environmental
Social
Physical:
Structured programmes
Irrelevant whether they are mainstream or not
Provide attention and guidance
Provide motivation
Allow adaptations
Type of activity
Fun
Purposeful
Incentive or reward
Team activity
(Barr and Shields 2011, Mahy et al 2010, Temple and Walkey 2007, Stanish et al 2006)
Family:
Parents:
Proactive
Provide opportunities
Enjoy sport themselves
Actively involved
Educating instructors
Siblings:
Motivating-encourage activity
Positive role-model
(Menear 2007, Barr and Shields 2011, Hutzler and Korenskv 2010)
Society:
Social Interaction
Primary reason to take part
Increase enjoyment
Provide encouragement
Opportunity for imitation
Reward for winning/partcipating
(Barr and Shields 2011, Hutzler and Korensky 2010)


BARRIERS
Physical:
Degree of functional limitation
Physical gap widening between them and peers
Communication difficulties
Hypotonicity
Weight and physique
Cardiac abnormalities
Reduced gross/fine motor skills
Lack of coordination
(Barr and Shields 2011, Menear 2007, Murphy and Carbone 2008, Wuang and Su 2011, Lotan 2007, Mahy et al 2010)
Personal factors
Behavioural:
Child preferences
Child's perceived self-competence
Lack of enjoyment
Frustration
Noncompliance
Lack of interest/motivation
(Murphy and Carbone 2008, Barr and Shields 2011,
Cognitive:
IQ/cognitive function – decreased IQ = decreased participation
Intellectual gap widening them and peers
(Rihtman 2009, Wuang and Su 2011, Barr and Shields 2011,Menear 2007)
Society:
Lack of social support
Lack of public awareness
Negative attitudes, stereotypes and Exclusive behaviour
Focus of coaches on winning
(Barr and Shields 2011, Murphy and Carbone 2008, Rimmer and Roland 2008, Templey and Walkey 2007)
Social
Environmental factors
Physical
Proximity to facilities
Lack of facilities
Lack of community programmes that are structured and suitable
Lack of trained staff
Transport
Finance
Lack of policy and political stand point on importance of PA
(Barr and Shields 2011, Murphy and Carbone 2008, Menear 2007, Mahy et al 2010, Templey and Walkey 2007)
Family:
Financial cost
Time available
Competing family responsibilities
lack of parent involvement
supervision requirements, safety concerns
overprotective parents
(Barr and Shields 2011, Murphy and Carbone 2008)

Assessment of PA (Stanish et al 2006):

- Use of multiple methods achieves the most accurate measurement although it may not be practical in a clinical environment
- Self-report
    - E.g. questionnaires, diaries
    - None have been validated in this population
    - Biased and unreliable due to reliance on the ability of individuals to recall PA
- Motion sensors/accelerometers
    - Allows objective calculation of activity frequency, intensity and duration
    - It has been successfully used in this population
    - It is expensive
- Pedometers
    - Not as sophisticated as accelerometers
    - Not able to measure general physical movement and cannot assess frequency and intensity of activity
    - Only expensive models can store data
    - Relativity high validity has been shown in this population

Useful Links

- Info on PA in adults with disability in Ireland: http://www.nda.ie/website/nda/cntmgmtnew.nsf/0/C6D0B0055C0B0FD880257878004C6E74/$File/PhysicalActivityHealthQualityofLife.pdf

- Guidelines to improve PA in youths (With and without a disability) http://health.gov/paguidelines/midcourse/PAG_Mid-course_Report.pdf http://health.gov/paguidelines/guidelines/default.aspx#toc

2.4.2. Employment and further education

There are limited studies available in this area and none were based on an Irish population.

In a longitudinal qualitative study by Carr (2008) of 54 parents of individuals with DS, 50% adults that were 30-35 years of age attended Social Education Centre full-time but at the age of 40 years 21% attended full-time and >50% attended part-time.

Very few of this sample had a job or had been prepared for a job. Only 15% had any work training. A total of 4/54 at 30 years, 3/54 at 35 years and 7/54 at age 40 years were working. From the age of 21 only 23% of the sample had ever worked and only 6% had had a paid job. However this was based on data obtained from a small sample of parents of people with DS. Melville et al (2008) included adults with DS and ID and found that the majority had a daytime occupation. However they did not give details as to whether this was paid employment, whether it was part-time or full-time, sheltered or competitive.

2.4.3. Housing

A qualitative study by Carr (2008) showed that at 21 years 71% still lived at home. This percentage gradually decreased with age – at 30 years of age 60% lived at home, 54% at 35 years old and 50% at 40 years old. At 40 years of age 35% lived in small group homes.

These percentage show that most continue to live at home although this study has a small sample and cannot be generalised to the wider population of adults with DS.

This study also showed that 25% would not be left in the house on their own and 50% would not be allowed out of the house on their own. These figures were directly related to maternal stress and perceived ability of the individual.

A larger cross-sectional study by Melville et al (2008) including a large sample of adults with ID (n=945) and DS (n=181) showed that the majority lived with a family carer or with paid support. A minority lived independently or in congregate housing.

2.4.4. Relationships and sexuality

Puberty (Van Cleve et al 2006)

- Begins at the same time as typically developing peers
- Sexual interest is similar to their peers
- Sexuality education is therefore important in adolescence and should include:
    - How to express physical affection in an appropriate manner
    - How to conform to family and societal standards for privacy and personal modesty
    - Discouragement of appropriate displays of affection in the community

- That they have the right to refuse inappropriate touching and how to refuse it

Fertility (Van Cleve et al 2006)

- Males have quite low fertility rates
- Women's fertility is not affected
- Offspring have a 50% chance of having DS

Relationships:

- A qualitative study by Carr (2008) reported that 50% of parents perceived that offspring with DS made friends easily. Over 80% had at least 1 friend but only a minority had a best friend. Social visits to friends declined with age and those with a non-disabled friend also declined with age.
- This study also reported that 25% had romantic relationships with a boy/girlfriend and although none were married 33% wanted to get married. 10/54 were in serious relationships and 2/54 had been engaged.

2.5 Key References

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- Mendonca, G.V., Pereira, F.D. and Fernhall, B. (2011) 'Effects of combined aerobic and resistance exercise in adults with and without Down syndrome', *Archives of Physical Medicine and Rehabilitation*, 92(1), 37-45
- Shields, N. and Taylor, N.F. (2010) 'A student led progressive resistance training program increases lower limb muscle strength in adolescents with Down syndrome', *Journal of Physiotherapy*, 56, 187-193 Barr, M. and Shields, N. (2011) 'Identifying the barriers and facilitators to participation in physical activity for children with Down Syndrome', *Journal of intellectual disability research*, 55(11), 1020-1033
- Skotko, B.G., Levine, S.P. and Goldstein, R. (2011) 'Having a brother or sister with Down Syndrome: Perspectives form siblings', *American journal of medical genetics,* 155(), 2348-2359

3. Duchenne Muscular Dystrophy (DMD)

3.1 Background and Diagnosis

3.1.1 Pathophysiology

Duchenne Muscular Dystrophy (DMD) is an X-linked recessive disorder for which there is currently no cure. Dystrophin is a protein present in muscle which connects muscle fibers to the surrounding extracellular matrix. Patients with DMD either have a total or partial absence of this protein which leads to the progressive loss of muscle strength, integrity and function. The disease first affects the proximal muscle groups and spreads distally. Respiratory, orthopaedic, and cardiac complications can manifest and lead to the inevitable mortality of the patient at the mean age of 19, (Bushby *et al* 2005).

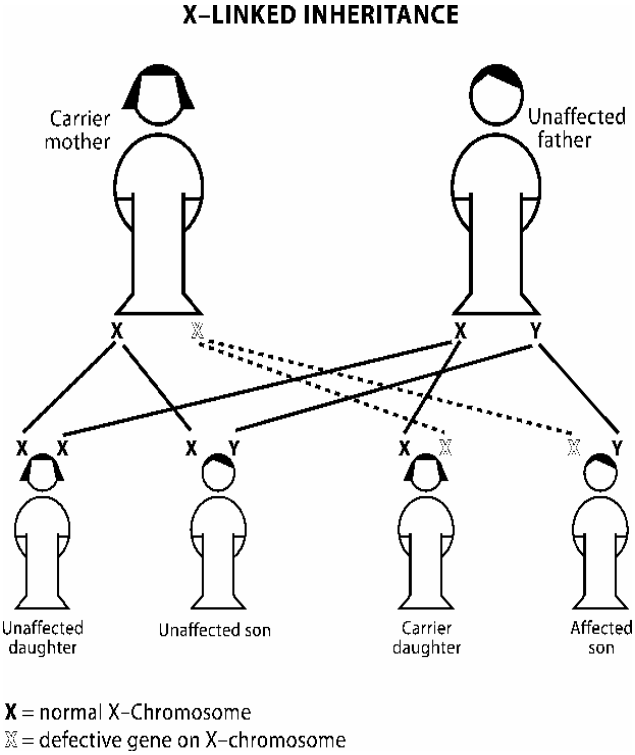
Due to the progressive nature of the disease, DMD is broken up into five different stages:

- Stage 1: Pre-symptomatic
- Stage 2: Early Ambulatory
- Stage 3: Late Ambulatory
- Stage 4: Early Non-Ambulatory
- Stage 5: Late Non-Ambulatory

3.1.2 Etiology

DMD is caused by a genetic mutation of the DNA which expresses the synthesis of dystrophin. Mutations lead to an absence of or defect in the protein dystrophin, which results in progressive muscle degeneration.

(Muscular Dystrophy Ireland 2013)



3.1.3 Epidemiology

DMD is the most common form of muscular dystrophy, occurring in about 1 in 3,500 males in the general population. However, research is equivocal on this figure with other studies suggesting an occurrence of 1 in every 2,400 males. The disease affects boys predominantly with cases of affected girls being rare – 1 in every 50,000,000, (Bushby *et al* 2010).

3.1.4 Clinical Features

Table 3.1.1 Clinical Features

Clinical Consideration	Description
Muscle weakness and contractures	Due to the absence of dystrophin, progressive muscle weakness is the main symptom of DMD. Proximal muscle groups are affected first in the lower limbs. This is irreversible but management strategies should be employed to maintain muscle power where necessary and to prolong the development of postural and spinal deformities. The overall goal is to promote function which in some cases requires reducing activity. In the late non-ambulatory stages, the disease pathology can lead to life-threatening impairments such as dysphagia and respiratory infections, Bushby *et al* (2010).
Learning and behavioural deficits	Children with chronic illness and/or physical disability have been shown to be at increased risk of learning and behavioural problems. Most boys with DMD present with specific mild cognitive deficits, characterized by poor verbal immediate memory and academic deficits. It is suggested that this is due to the physical limitations associated with DMD and also possibly due to the absence of dystrophin in the brain, Hilton *et al* (2006); Bushby *et al* (2010).
Cardiovascular function	Cardiomyopathy and/or cardiac arrhythmias are common features of DMD and almost always present. However, they develop asymptomatically during the early ambulatory stages of the disease. If left untreated, heart failure can ensue increasing morbidity and risk of early mortality. Assessment and close surveillance is vital from the time of diagnosis so that it can be treated promptly, Bushby *et al* (2010).

Respiratory function	Respiratory problems in DMD tend to be very predictable and correlate with overall muscle strength. Boys who lose ambulation early are likely to require ventilation sooner than those who walk longer. Essentially respiratory function during the ambulant stages is normal and management issues usually don't arise until the non-ambulant stages. However, vigilance and monitoring is advised, Bushy *et al* (2005); Bushby *et al* (2010).

3.1.5 Diagnosis

DMD is a difficult condition to diagnose during the first few years (presymptomatic stage) with the average age of diagnosis being four and a half, (Bushby *et al* 2005). The reason for this is that often the visible symptoms of DMD cannot be observed until the young boy starts to become more ambulant. Furthermore, it can be difficult to distinguish DMD from other neuromuscular diseases such as Becker's Muscular Dystrophy (BMD) which is a milder form of DMD.

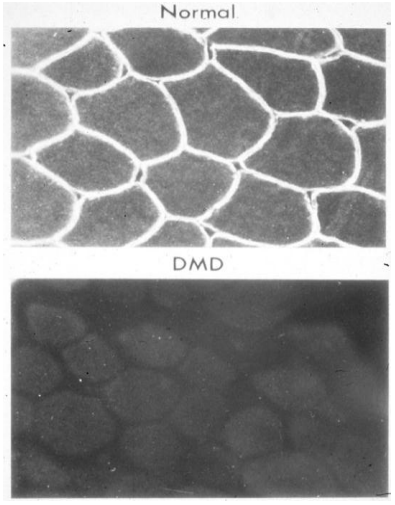
Bushby *et al* (2010)

3.1.6 Clinical Examination

The clinician must observe the child performing functions such as running, jumping, climbing stairs and getting up from the floor. Signs of proximal weakness are symptomatic of DMD. Such signs are *Gowers' sign*, waddling gait, toe-walking, frequent falls, and the inability to climb stairs. The child is also likely to have a history of developmental delay with regard to attaining his milestones. Furthermore, there may be a delay in language development.

In some cases, there may be a known family history of DMD. In such situations, if any suspicion of abnormal muscle function arises, the clinician should refer the child immediately for DMD screening. If there is no family history of DMD and the child is not walking by >16-18 months, the clinician should refer the child for DMD screening, Bushby *et al* (2010).

Table 3.1.2 DMD Screening Process

Test	Description
Creatine Kinase Concentration	In DMD, there is a massive elevation in the serum concentration of creatine kinase. This enzyme is secreted after muscle damage. A positive result for this test however is non-specific for diagnosing DMD. Nevertheless, it should prompt urgent specialist referral for confirmation diagnostics, Bushby *et al* (2005).
Muscle Biopsy (1) **Muscle Biopsy (2)**	The muscle biopsy shows abnormalities typical of muscular dystrophy such as necrosis and attempted regeneration of individual muscle fibres, increased variability of muscle fibre diameter, ISNO (2012). To further investigate the clinical diagnosis, immune-histochemical analysis of the muscle biopsy is usually performed, ISNO (2012). If this shows complete absence or less than 5% of muscle fibres staining positive for the presence of dystrophin, further genetic analysis is performed to confirm diagnosis of DMD Bakker et al, (1997). If there is only a partial loss of dystrophin, this may be a milder form of muscular dystrophy such as Becker's Muscular Dystrophy.  Figure 3.1.1. Muscle biopsy Top: normal muscle showing dystrophin round the fibres. Bottom: dystrophic muscle with absent dystrophin

Genetic Testing	A positive genetic test reveals a deletion of the dystrophin gene (70%), duplication of the gene (6%) or in the remaining cases, a point mutation. If one of these observations is found then no further testing is required. However, a negative result does not always exclude the disease. It is very important to recognise what tests are offered by a particular laboratory, and what are their limitations, Bushby *et al* (2010).

3.1.7 Genetic Counselling and Carrier Testing

When the genetic mutation causing DMD arises by chance in the boy, this is called spontaneous mutation. In other cases, the mutation has been passed on by the boy's mother, the "carrier". This means she could have passed the genetic mutation on to previous children, and could pass it onto future children too. The boys she passes it on to will be affected by DMD, while the girls will be carriers. If the mother is tested and is found to have the mutation, she can make informed decisions about future pregnancies, and her female relatives (sisters, aunts, daughters) can also be tested to see if they are also at risk of having a boy with DMD. Even when a woman is not a carrier, there is a small risk to future pregnancies because the mutation may occur in her ova or egg cells. This is called "germ line mosaicism".

Bushby *et al* (2010).

3.1.8 Prognosis

The progressive nature of the disease results in the young boy needing a wheelchair at the average age of 9, the later development of cardio-respiratory complications, and the inevitable mortality at the mean age of 19. This is a general perspective. However, with improved care and developing research, longevity is increasing with some patients living into their 30's.

3.2 Health Management of DMD

The following information has been sourced from two key sources; Bushby *et al* (2010) Part 1 and Part 2. Their research provides a comprehensive, in-depth, expert analysis of the literature surrounding MDT management throughout each stage of DMD. The level of evidence for various treatments and procedures is also described. Although no accredited management guidelines specific to DMD were found in forming this short-course, there are guidelines based on expert opinion that were formulated prior to the Bushby et al (2010) trials. These guidelines are named 'Standards of care for Duchenne muscular dystrophy Brief TREAT-NMD recommendations' and accessed at http://www.parentprojectmd.org.

The goal of DMD management is to optimise patient independence and quality of life through planning, maintenance, prolonging complications and palliative care, Bushby *et al* (2010).

3.2.1 Pre-Symptomatic Stage

Given the predictable nature of DMD, anticipatory and preventive measures should be strategically set in place by each member of the multidisciplinary team (MDT). MDT management is an on-going process for a patient with DMD. However, as stated above the average age of diagnosis is 5. Therefore, in most cases, MDT management and planning will not be fully implemented until this point, Bushby *et al* (2010). Before this point in time, the child is more than likely in the pre-symptomatic stage displaying mild symptoms of motor delay and /or mild proximal muscle weakness. It is important to remember that DMD does not present exactly the same for every child at certain ages. While most young boys with DMD tend to follow a similar age-related disease progression, in some cases the disease can manifest and progress more rapidly reaching the non-ambulatory stages sooner than predicted. The DMD patient must be managed in relation to what stage disease he is at and how he presents symptomatically, (Mary Delaney – Paediatric Physiotherapist).

Early Intervention (EI)

Despite whether or not the patient is diagnosed with DMD, he is likely to present with some element of developmental delay, Bushby *et al* (2010). Thus, he may require Early Intervention (EI) assessment and treatment involving an MDT approach. This approach aims to identify the child's needs. Based on this assessment and the severity of the child's condition, the child's needs can be specifically managed by the necessary teams.

3.2.2 Neuromuscular Management

Assessment

From the point of diagnosis onwards, the young child should be assessed every 6 months by a specialist physician to monitor the disease progression and to identify any further neuromuscular abnormalities that may require further investigation. The physician can then decide on the optimal choice of pharmacological treatment to help preserve and/or enhance neuromuscular function. Dosage adjustments can be made based on assessment findings as is necessary.

Steroid

Glucocorticoid steroids (GCS's), are the mainstay of medical-neuromuscular treatment in DMD. They are currently the only drugs known to slow the decline in muscle strength and motor function in DMD. *Prednisone* and *deflazacort* are the two types of steroids that are mainly used in DMD. *Deflazacort* is prescribed if the patient presents with pre-existing weight and/or behavioural issues. The main benefits of steroid use in DMD are:

- they help the child walk independently for longer
- they help minimise and stabilise pulmonary, cardiac and orthopaedic problems i.e. cardiomyopathy and scoliosis
- they enhance participation

(Cochrane Review Manzur *et al* 2008)

(Moxley *et al* 2005)

Despite these benefits, there are a number of significant side effects with the long term use of steroids. Therefore, structures must be put in place to help minimize these side-effects. For a full description of these side effects, refer to the full booklet located on SULIS.

The physician should discuss the initiation of GCS's with parents/caregivers prior to being prescribed. Consideration should be given to the patient's age, function and pre-existing risk factors before commencing steroid treatment. Furthermore, certain immunisations are mandatory before initiating steroid treatment due to the heightened risk of infection while on the course. In general, initiation of steroid treatment commences at a stage known as the "plateau phase". This is when the boy's motor skills have stopped improving, but have not yet started to get worse. This phase can be different for each child but on average it is around the age 4-6yrs. Boys who start steroid treatment during the ambulatory stages usually continue this treatment during the non-ambulatory stages with careful alterations to dosage often necessary.

Initiation of steroid treatment is not recommended for a child whose motor skills are still improving and especially those under the age of 2yrs who are still gaining the basic motor skills. Initiation of steroid use in the non-ambulatory stage of disease is a matter of individual decision and should be discussed with the physician. The patient, parents/caregivers and clinicians need to decide whether the benefits outweigh the risks or not. The benefit of GCS's at this stage is to maintain upper limb function and prevent scoliosis. However, due to reduced mobility, the side effects of treatment may become more pronounced.

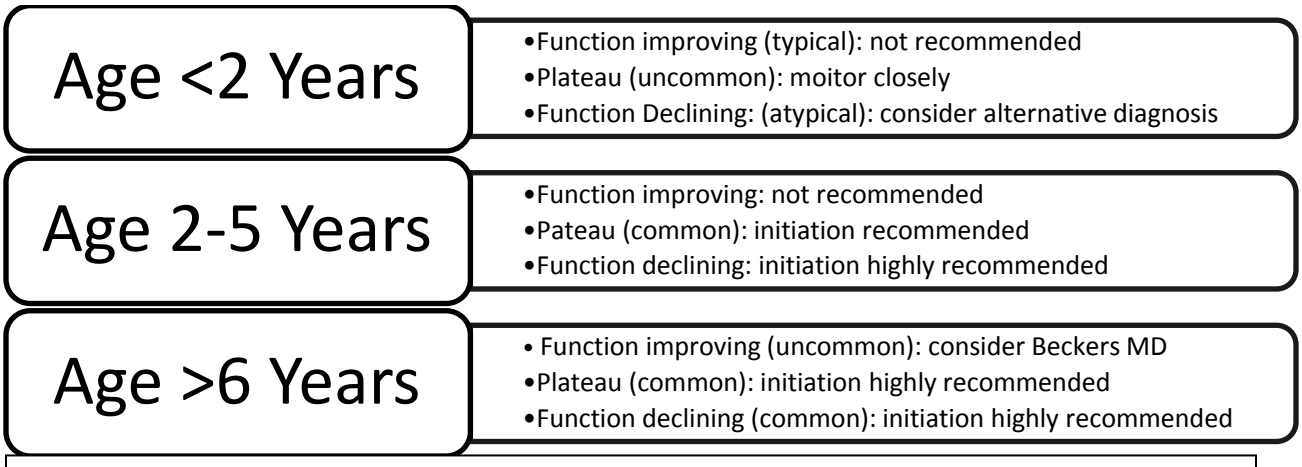


Figure 3.2.1: Recommended Times to Initiate Steroid Treatment based on Functional Status

Currently, there is a considerable amount of ongoing research in the area of DMD and pharmacology. New drugs are being produced to help slow the progression of the disease. However, there is insufficient evidence for their use at present, Bushby (2010).

Physiotherapy and Occupational Therapy

The child with DMD should be assessed by a physiotherapist and/or Occupational Therapist (OT) every 4months approximately. Information regarding exercise and the role of the physiotherapist in DMD is provided below.

The OT may be involved in the early ambulatory phase in facilitating the child's fine motor development – clinician opinion. The OT is also vital in prescribing mobility aids. During the ambulatory stages, depending on the child's level of functioning, a scooter or stroller may be used for mobility purposes over long distances to conserve strength. As the disease progresses and the child enters the late ambulatory stage, wheelchair prescription is advised sooner rather than later. A balance between independent functioning and energy conservation is recommended. During the non-ambulatory stages, seating is an important consideration. The OT should regularly assess and provide seating equipment that promotes good posture and is comfortable.

3.2.3 Cardiovascular Management

Cardiomyopathy is an almost universal complication and a major source of morbidity and mortality in patients with DMD. The point of onset of cardiomyopathy is poorly understood but what research has shown is that the disease is present long before symptoms become evident, Bushby *et al* (2010). During the early stages of DMD, the disorder commonly progresses asymptomatically. However, if it is not detected until the later non-ambulatory stages of DMD, the heart may be damaged to a point of heart failure (HF) for which there is a poor prognosis. Furthermore, symptoms of HF such as fatigue, weight loss, vomiting, abdominal pain, sleep disturbance and inability to carry out ADL's are often disguised under the musculoskeletal limitations of DMD. This means that often patients must commence preventative treatment long before symptoms develop due to the early and progressive nature of the heart disease.

More information on cardiomyopathy can be found on the Irish Heart Foundation webpage through the following link: http://www.irishheart.ie/iopen24/cardiomyopathy-t-55.html

Assessment and monitoring

Cardiac assessment and monitoring is recommended from the point of diagnosis and then every two years up to age 10, Bushby *et al* (2010). This includes an electrocardiogram (ECG) and an

echocardiogram. This provides a baseline measure for the cardiac specialist to use to help identify the developing abnormalities and the need for intervention after future assessments. Common assessment findings in patients with cardiomyopathy include left ventricle hypertrophy, patent ductus arteriosus, artrial and/or ventricular septal defects and sinus tachycardia.

If any of these abnormalities are identified, increased surveillance and pharmacological treatment is warranted irrespective of age. However, during the ambulatory stages of the disease, cardiac problems are unlikely to manifest. It is estimated that 20-30% of DMD boys have left ventricular impairment by the age of 10. From the age of 10 years onwards, as the patient becomes less mobile, surveillance of abnormalities should become even more vigilant. Moreover, patients on GCS's require additional monitoring due to side-effects.

Treatment

First-line treatment for patients with DMD is ACE-inhibitors. ACE-inhibitors work by promoting vasodilation of the blood vessels and by reducing the activity of the sympathetic nervous system. This overall improves haemodynamics which benefits the patient by promoting left ventricular function and slowing disease progression, Davies *et al* (2000).

NHS guidelines 2010 recommend *enalapril, lisinopril,* and *ramipril* for the treatment of heart failure. Bushby e*t al* (2010), recommend following these standardised guidelines in the management of heart failure in DMD. These drugs have the most medical evidence to support their use, and doctors generally have more experience using them. Some research supports the use of ACE inhibitors to treat cardiomyopathy prior to any signs of abnormal functioning. Despite this finding, further research is required before recommendations can be made.

3.2.4 Gastro-Intestinal Management

Nutrition

At any stage of DMD, the patient may be at risk of under-nutrition or being overweight due to multiple reasons, e.g. reduced physical activity, swallowing impairment, muscle atrophy, GCS's side effects, growth etc. The child's age related body mass index (BMI) should be monitored and kept between the 10th and 85th percentile. If the child is outside these limits he should be

referred onto a dietician who will assess the child's calorie intake, energy output, and provide information on diet and sources of nutrients. Also, patients who are on long-term steroid use are at higher risk of fractures. Calcium supplementation can be prescribed to help avoid such complications. The child should be referred for a nutritional/dietetic assessment by any member of the MDT at any of the following stages:

- At diagnosis
- At initiation of GCS's
- If the patient less than the 10th age percentile or over the >95th age percentile BMI
- If there has been unintentional weight loss or gain
- If major surgery is planned
- If the patient is chronically constipated
- If dysphagia is present (Bushby *et al* Part 2)

Swallowing

There is an increased the risk of dysphagia in patients with DMD. This increases the risk of severe weight loss. Furthermore, the patient is at a heightened risk of aspiration. Vigilant surveillance is therefore critical, especially during the later stages of the disease to identify any indication of dysphagia. Such indicators are:

- Unintentional weight loss of 10% or more
- Insufficient weight gain in growing children
- Prolonged meal times (>30mins)
- Meal times accompanied by fatigue, drooling, coughing or choking

Any of these indicators warrant a clinical swallowing examination. If swallowing difficulties are identified, the patient should be referred onto the speech and language therapist who will devise an individualised therapeutic programme to maintain good swallow and/or develop appropriate compensatory strategies.

As the disease progresses and the patient enters the final stage, swallowing difficulties may be accentuated due to weakened chewing and swallowing muscles. When it is no longer possible to maintain weight and hydration by oral means, gastric-tube placement (gastrostomy) is indicated.

More information can be found in the Bushby trials (Bushby *et al* 2010).

Gastro-Intestinal Problems

Constipation and gastro-oesophageal reflux disorder (GORD) are the two most common gastrointestinal conditions seen in children with DMD in clinical practice. Constipation occurs most frequently in later stage DMD patients after surgery due to the effects of anaesthesia, inactivity and diet. However, this complication can often arise during the non-ambulatory stages due to decreased mobility. GORD can occur in patients with DMD due to the side-effect of steroids and is more commonly found in patients who are overweight/obese.

Speech and Language

Delayed acquisition of early language milestones is common in boys who have DMD. This is due to an:

- impaired short-term memory
- impaired phonological processing
- impaired IQ
- a specific learning disorder

Such impairments lead to a difficulty in language acquisition and skill throughout the boy's childhood. To help children with DMD learn and develop the necessary language skills, and to overcome language deficits, they can be referred to a speech and language therapist (SLT). The SLT will assess and treat children based on their impairments. Oral motor exercises and articulation therapy can be of benefit to young DMD patients with hypotonia and also to older DMD patients who have deteriorating oral muscle strength. In the later stage of DMD, it can often become difficult to understand the patient's speech due to impaired respiratory support for speech and vocal intensity. In such cases, compensatory strategies, voice exercises, and speech amplifications are appropriate interventions (Bushby *et al* 2010 Part 2).

3.2.5 Orthopaedic Management

Orthopaedic Management at each stage of DMD

- In the early ambulatory stage orthopaedic surgery is rarely necessary
- In the late ambulatory stage, surgery may be considered for achilles tendon contractures
- In the non-ambulatory stage, scoliosis should be monitored. Intervention with spinal fusion may be considered. Also intervention for foot position may be considered to improve wheelchair positioning on footrests, to alleviate pain and pressure and allow shoes to be worn (Bushby *et al* 2010 Part 1)

Joint Contractures

Physiotherapy is the first line treatment to help maintain muscle length and integrity and to prevent the development of contractures. Range of motion exercises, splinting and other interventions should be prescribed to every child from the point of diagnosis onwards. Some patients will require more intensive treatment than others.

- Reasons for ↓ muscle extensibility and contractures.
- Importance of maintaining ROM

(Bushby et al 2010 Part 2)

Stretching

Stretching should be carried out at least 4-6 days per week, at home, in school and during physiotherapy. Effective stretching requires combined interventions including:

- Active stretching
- Active-assisted stretching
- Passive Stretching
- Prolonged muscle elongation using positioning
- Splinting
- Orthoses
- Standing Devices

(Bushby *et al* 2010 Part 2)

Orthoses

- Orthoses and standing devices can also help reduce contractures. Refer to Bushby *et al* (2010) Part 2.

- A systematic review by Bakker *et al* (2000) looked at the effects of KAFO's in DMD. The review concluded it appears that knee ankle foot orthotics (KAFO) can prolong assisted walking and standing, however it is unclear if it can prolong functional ambulation. The boys that gained the most from the KAFOs have a low rate of severity, are capable of enduring an operation and are motivated. The overall quality of the trials used in the review is poor.

Standing Devices

Necessary for late ambulatory and early non-ambulatory stages provided there are no contractures or mild lower limb contractures (Bushby *et al* 2010 Part 2)

Surgical Intervention for Lower Limb Contractures

Due to the inevitable progressive nature of DMD, contractures may develop despite best efforts. To help preserve functional ambulation during the early stages of the disease, surgical intervention is an option. The joints most amendable with surgical correction include the ankle and the knee. Although flexion contractures are common at the hip, surgical outcomes are generally poor. Surgical release or lengthening of iliopsoas may further weaken the muscle and decrease overall function of the patient. The most common types of surgical interventions for the lower limb include:

- Tendo-Achilles lengthening
- Hamstring tendon lengthening
- Anterior hip-muscle release
- Excision of iliotibial band

In general, surgery is performed during the ambulatory phases of the disease. Surgical intervention depends on patient specific circumstances. The effect of anaesthetic must be considered (Bushby *et al* 2010 Part 2).

A Cochrane review by Brittle *et al* (2009) reviewed rehab for foot drop (weakness or contracture at the ankle) resulting from neuromuscular disease. Of the four RCT's included in the review, one RCT involved the DMD population and looked at Achilles tendon Surgery. The surgery had no significant effect on walking ability in children between 4-6years. The review concluded there is some evidence that early surgery is not effective in people with DMD in terms of walking speed, muscle strength or other measures of functional 'motor ability' at one, two or nine years after surgery (Manzur 1992 as cited in Brittle *et al* 2009).

Assistive/Adaptive Devices for Function
Refer to (Bushby *et al* 2010 Part 2).

Spinal Management
Scoliosis is common in Duchenne muscular dystrophy (Baxter et al 2007). Children with DMD who are not undergoing glucocorticoid steroid (GCS) treatment have a 90% chance of developing significant progressive scoliosis, Smith et al (1989). Even with GCS therapy, there is no guarantee that the child will not develop scoliosis. Some argue that steroids merely delay its onset. It is noteworthy that steroids may reduce the risk of scoliosis. However they increase the risk of vertebral fracture (Bushby *et al* 2010 Part 2).

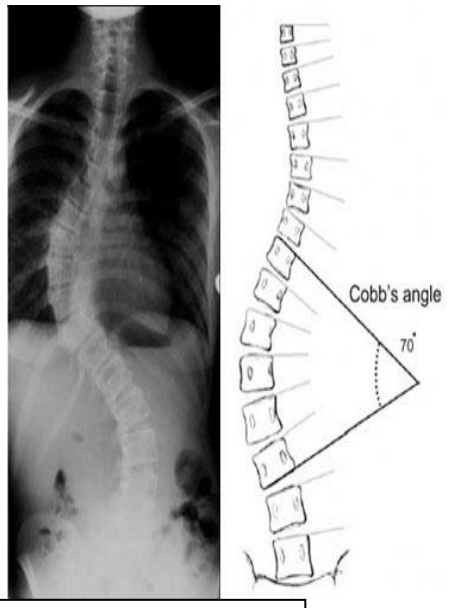


Figure 3.2.2 X-ray of scoliosis

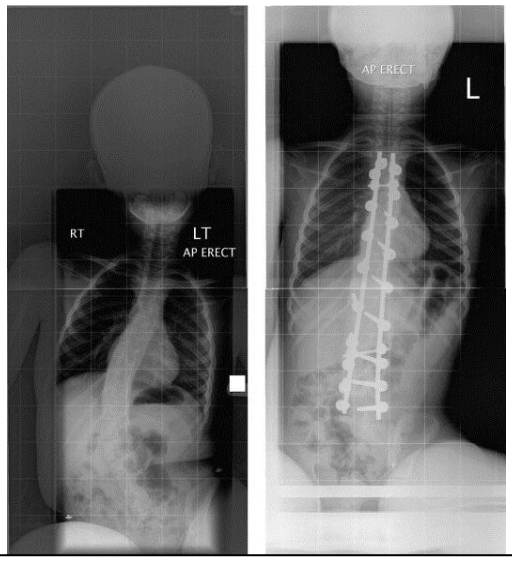


Figure 3.2.3 Scoliosis pre and post-op

The orthopaedic surgeon should closely monitor the posture and spinal alignment of the child throughout the ambulant stages of the disease. This is achieved by clinical observation. If there is suspicion of a scoliosis, an X-ray may be required. Vigilance is especially required when the child is rapidly growing. Once the child enters the non-ambulant stage, a sitting anteroposterior full-spine X-ray is required as a baseline assessment of the spine. Annual follow up X-rays can then be compared to examine the development or progression of a scoliosis. For more details on spinal management, including spinal surgery refer to the following reviews: Baxter *et al* (2007), Bushby *et al* (2010) Part 2.

The Cochrane review by Baxter *et al* (2010) concluded there is no high quality evidence for scoliosis surgery in DMD, as there are no RCTs available.

Bone Health Management
Consensus statements on bone health in DMD: Davie *et al* 2005;Bachrach *et al* 2005.

Why is there reduced bone health?

- Reduced physical activity
- Weak Muscles (Bushby *et al* 2010 Part 2)
- Glucocorticoid Therapy (Although steroids reduce scoliosis risk (or delay onset), they increase the risk of vertebral fractures (Bothwell *et al* 2003)

For further details on bone health management and fracture management refer to Bushby *et al* (2010) Part 2.

3.2.6. Respiratory Management

The evidence for much of current practice is weak and is based predominantly on observational trials (Aniapravan *et al* 2012). The following information has been collaborated from the British and American Thoracic Society guidelines, whereby existing evidence was used and where lacking, expert consensus opinion was formulated. Key information was also

sourced from a review initiated by the US Centres for Disease Control and Prevention. The review involved the DMD Care Considerations Working Group evaluating assessments and treatments in DMD, in order to develop care recommendations and improve practice (Bushby *et al* 2012 Part 1&2).

Risk of Respiratory Complication

In the ambulatory stage of DMD, there is a low risk of respiratory problems. However it is recommended to monitor respiratory status (measure FVC at least annually) as the condition is progressive (Bushby *et al* 2010 Part 1). Once the patient becomes non-ambulant, there is an increasing risk of respiratory impairment, triggering the need for pulmonary assessment. As the individual enters the late non-ambulatory stage he/she is at high risk of respiratory impairment, therefore requires respiratory investigation and intervention.

The most common reason for unplanned hospital admission is acute respiratory failure with associated respiratory infection. Chronic respiratory failure is frequently the cause of death. Through a multi-disciplinary approach to assessment and treatment, the frequency of hospital admission will be reduced and life expectancy will improve (Aniapravan *et al* 2012).

Complications

In DMD there is a risk of respiratory complications as the condition progresses. This is secondary to a reduction in respiratory muscle strength. The patient with non-ambulatory stage DMD is at risk of reduced airway clearance. The individual retains mucociliary clearance while losing cough clearance (ATS 2004). Other respiratory complications include nocturnal hypoventilation, sleep disordered breathing and eventually daytime respiratory failure (Bushby *et al* 2010 Part 2).

Stages of Respiratory Impairment

1. Reduced airway clearance secondary to ineffective cough.
2. Nocturnal Hypoventilation
3. Inadequate Ventilation in Daytime

(Finder 2009)

Frequency of Assessment

The American Thoracic Society (ATS) consensus statement in 2004 recommends early (presymptomatic stage) assessment by a respiratory specialist and ongoing checks of respiratory status. Twice yearly screening is advised:

- Once in the non-ambulatory stage or
- after 12years of age or if
- FVC is <80% predicted.

Once mechanical ventilation or assisted cough is being used, screens should be carried out every 3-6months.

The British Thoracic Society guidelines recommend Vital Capacity (VC) should be measured in all patients who are capable of using spirometry (Level C Evidence) and cough peak flow should be used to assess secretion clearance in those over 12 years.

Sleep disordered breathing should be assessed at least annually if:

- non-ambulant
- VC is <60% predicted
- symptoms of obstructive sleep apnoea
- hypoventilation
- diaphragmatic weakness
- rigid spine syndromes

Bushby *et al* (2010) Part 2 provides further information on frequency of assessment and recommended respiratory measurements at each stage of DMD.

Interventions

- The basic aims of respiratory care of an individual with DMD (and for patients with all forms of weakness) are to encourage airway clearance and support breathing (ATS 2004).

- In the literature assisted cough and nocturnal ventilation have been shown to improve survival in DMD (Bushby *et al* 2010 Part 2).

Assisted Cough

- Manually assisted cough is supported by both the British (Level C evidence) and American Thoracic Society guidelines (Aniapravan *et al* 2012; ATS 2004).
- Mechanical insufflation-exsufflation (MI-E) is strongly supported for patients with reduced cough clearance (ATS 2004). The BTS guidelines advise MI-E should be considered in very weak children, with loss of bulbar function, or in those who cannot co-operate with manual cough techniques (Aniapravan *et al* 2012). MI-E is an alternative airway clearance technique (ACT) aiming to reduce deterioration and the need for intubation and mechanical ventilation (Aniapravan *et al* 2012).

Assisted Ventilation

- Assisted ventilation via tracheostomy can prolong survival; however non-invasive ventilation is advocated (Bushby *et al* 2010 Part 2).
- NIV should be offered where possible and invasive options should be used when those NIV options are not successful (Aniapravan *et al* 2012; ATS 2004).
- NIV should be first line treatment in acute respiratory failure (Aniapravan *et al* 2012).
- Indications for Nocturnal Ventilation:
    - Signs or Symptoms of Hypoventilation
    - Baseline SaO2 <95% and/or PCO2 >6kPa while awake (Bushby *et al* 2010 Part 2)

- Indications for Daytime Ventilator Support:
    - Daytime Hypercapnia (Exhaled CO2 >6.7kPa) and/or
    - Haemoglobin saturation at <92% while awake
    - Inability to speak a full sentence without breathlessness.

    (Aniapravan *et al* 2012; ATS 2004, Bushby *et al* 2010 (Part2)

- Indications for Invasive Support:
    - Patient/Family preference
    - Bulbar weakness (weakness of tongue, lips, phaynx and larynx).

    (Aniapravan *et al* 2012; ATS 2004)

- ICU: Criteria for Extubation:
    - Minimal airway secretions
    - Effective use of ACT e.g. MI-E
    - SaO2 >94% on room air for >12hrs.
    - Continuous NIV should be used straight after extubation.

*Note: See Bushby *et al* (2010) Part 2 for further information on interventions.

Evidence for airway clearance techniques provided by BTS

- Independent ACTs such as the use of deep inspiration and FET (Huff or Cough) are ineffective in children with significant neuromuscular disease as they do not have the ability to take a deep breath or cough effectively.
- Other ACTs should be used. Expert opinion strongly advises respiratory physiotherapy during acute pulmonary infection. There are currently no controlled trials, as use of a placebo would be unethical.
- The BTS Guidelines recommend ACTs during pulmonary infection when SaO2 <95% on room air, Level D evidence.
- Consider oscillatory techniques (high-frequency chest wall oscillation and intrapulmonary percussive ventilation) in children who have difficulty mobilising sputum or who have atelectasis, even after the use of other ACTs, Level D evidence (Aniapravan *et al* 2012)
- Air- stacking and manually assisted cough are effective in improving cough effectiveness, Level C evidence (Aniapravan *et al* 2012)

- ACTs which improve cough efficiency as part of home treatment which involves NIV results in reduced hospitalisation for respiratory exacerbation and increases survival, Level 3 Evidence (Aniapravan *et al* 2012)
- Choosing an ACT depends on:
    - Child's ability to co-operate with ACT
    - Extent of child's weakness, Level 3 evidence (Aniapravan *et al* 2012)

Respiratory Muscle Training

- Respiratory muscle training improves respiratory muscle strength and endurance in the healthy population. In people with DMD, respiratory muscle training may be introduced with the aim of preserving or increasing ventilation capacity and decreasing risk of exacerbation by improving cough effectiveness (Aniapravan *et al* 2012).
- The ATS consensus statement (2004) did not support respiratory muscle training secondary to conflicting data in the literature. There was an issue that muscle training might actually harm the individual (due to deficiency of nitric oxide).
- Recent British Thoracic Society (BTS) guidelines (2012) state respiratory muscle training can improve respiratory muscle strength and endurance in children and young adults with DMD (Evidence Level 1-) (Aniapravan *et al* 2012).
- Two longer-term (9 and 24 months duration) observational studies reviewed by the BTS guidelines showed increases in respiratory muscle strength and endurance while training was maintained. There was no evidence of increased muscle damage caused by training in either study (Aniapravan *et al* 2012).
- No studies have assessed effects on clinically important outcomes of ability to maintain breathing function over time, or decrease respiratory exacerbation.

Considerations during established infection

- Take caution introducing oxygen therapy, as supplemental oxygen can improve hypoxaemia, but hide the underlying cause e.g. atelectasis or hypoventilation. It is

possible that supplemental oxygen will undermine central respiratory drive and worsen hypercapnia.

- NIV and assisted cough (manual and mechanical) are essential if hypoxaemia is secondary to reduced airway clearance, hypoventilation and/or atelectasis (Aniapravan *et al* 2012; Bushby 2010 *et al* Part 2).
- The BTS guidelines recommend ACTs should be used during respiratory infection if SaO2 <95% on room air (Aniapravan *et al* 2012).

Post-op Respiratory Considerations for DMD

- Aim to prevent atelectasis, pneumonia and post-op endotracheal extubation failure.
- NIV and assisted cough should be considered post-op if pre-operative PFT's were below normal, secondary to significant respiratory muscle weakness. Pre-op practice and post-op use of NIV is highly recommended for individuals with a baseline FVC of < 50%predicted and essential with FVC <30% predicted.
- Incentive spirometry is not indicated secondary to possible lack of efficacy in people with respiratory muscle weakness (Bushy *et al* 2010 Part 2, Aniapravan *et al* 2012)

Effect of steroids on respiratory muscle function (BTS Guidelines)

Non-randomised trials show a long–term improvement on respiratory status. Biggar *et al* 2006 (as cited in Aniapraven *et al* 2012) reported significant difference in VC between boys prescribed deflazacort and boys not receiving at ages 10, 15 and 18. By the age of 18, 46% of the control group and none of the treatment group needed nocturnal NIV (Aniapravan *et al* 2012).

3.2.7 Exercise in DMD

Physical activity Levels

- In muscle disease, there are a number of factors leading to reduced physical activity and a sedentary lifestyle:
  - Muscle weakness
  - Fatigue
  - Pain
  - Difficulty exercising (McDonald 2002 as cited in Guerts *et al* 2010)

- Frequency of Falls(Requiring help to stand up) and Fear of Falling (de Groot *et al* 2010)
- Cardiorespiratory morbidity and progressive joint contractures and deformity (Carter *et al* 2012).

- Physical inactivity negatively impacts on quality of life and health (McDonald 2002 as cited in Guerts *et al* 2010).

Recommendations for Exercise

- Currently there is limited available research on recommendations for exercise in DMD (Bushby *et al* 2010 Part 2). Studies have not yet resolved if exercise is beneficial in DMD (de Groot *et al* 2010). Although some recommendations are available, specific prescription guidelines (FITT principle) are unclear (de Groot *et al* 2010)
- Submaximal functional exercise is essential to prevent atrophy secondary to disuse particularly in individuals in the ambulatory stage of DMD (Bushby *et al* 2010 Part 2).
- Exercise should include swimming-pool exercise and land-based exercise. Swimming is highly recommended, which may improve aerobic fitness (Bushby *et al* 2010 Part 2) and improve or maintain mobility and strength (Ciafaloni and Moxley 2008)
- Low-resistance training and upper limb function exercise may also benefit (Bushby *et al* 2010 Part 2).
- Studies of strength training in DMD subjects have demonstrated preservation of strength or even mild improvement over the period of the trial. But trials are limited by use of non-quantitative measures, lack of control groups, and use of opposing limb as a control without factoring in the impact of cross-training (Carter *et al* 2012) The use of the opposing limb as a control also makes it difficult to assess improvement in function.

Muscle Injury

- Post exercise reports of overexertion and weakness in people with muscle disease have resulted in a cautious approach to training (Geurts *et al* 2011).

- High resistance training and eccentric training are contra-indicated across the lifespan secondary to concerns about contraction-induced muscle injury (Bushby *et al* 2010 Part 2).
- Substantial muscular pain or myoglobinuria within 24hr of a specific activity is indicative of overexertion and contraction induced injury, therefore the exercise requires modification (Bushby *et al* 2010 Part 2).

Systematic Review on exercise in Muscle Disease

- A Cochrane review looked at resistance training and aerobic training in muscle disease. The review was unable to provide evidence based guidelines for DMD, due to scarcity of RCT's. The review concluded that for myotonic dystrophy, fascioscapulohumeral muscular dystrophy and mitochondrial myopathy, individuals can be advised that 'normal' activity in sports and work seems not to damage their muscles, however there is insufficient evidence that it provides benefit (Geurts *et al* 2010). The above conclusion is not generalisable to DMD, secondary to varying pathophysiology in the different types of muscle disease and therefore potentially different responses to training.
- The ACSM guidelines (Pollock 1998 as cited in Guerts *et al* 2011) for exercise in healthy individuals were used by most of the included and excluded trials in the Cochrane review by (Guerts *et al* 2010)

Need for Further Research

- More research is required to determine whether strength training and aerobic exercise are advantageous in DMD and to determine optimum exercise parameters (Geurts *et al* 2010). There is a requirement for further studies to determine safe and potentially beneficial types and amounts of activity and exercise as well as to identify possibly harmful types and amounts of activity and exercise (Carter *et al* 2012)
- The purpose of exercise in increasing quality of life in DMD needs further research (Geurts *et al* 2010).
- A study protocol published in 2010 for the 'No Use is Disuse' study may help provide evidence for exercise in DMD. It is the first study involving people with DMD to look at whether low-intensity physical exercise is advantageous in maintaining muscle endurance and functional abilities (de Groot *et al* 2010). Awaiting results.

3.3 Psychological

3.3.1 Psychosocial Aspect (Adolescence and Adulthood)

As part of multi-disciplinary team management, the psychosocial wellbeing of people with DMD must be addressed (Bushby *et al* 2010 Part 1). Up to 30-50% of people with DMD have psychosocial issues including general emotional or behavioural disturbance, symptoms of depression and social problems (Aldenkamp *et al* 2009). There are a number of factors that can affect psychosocial health including biological factors (lack of dystrophin which affects brain development and function) (Donders and Taneja 2009), social factors, emotional factors and treatment factors (e.g. glucocorticoids). Even though most of the psychosocial factors are not specific to the DMD population, patients with DMD are at increased risk of developing psychosocial issues (Bushby *et al* 2010 Part 1).

Psychosocial Issues

- Issues with social functioning (Donders and Taneja 2009) could be secondary to deficits in cognition, while physical limitations may result in social withdrawal, social isolation and reduced access to social activities (Bushby *et al* 2010 Part 1) .
- Speech and language deficits and cognitive delays are well documented in DMD. There is also more risk of autism, attention deficit hyperactivity disorder and obsessive compulsive disorder in DMD, which may impact psychosocial function.
- Issues with emotional adjustment and depression.
- Issues of anxiety exacerbated by cognitive deficits.
- Psychosocial issues related to independence and vocational planning that is not being regularly assessed (Rahbek *et al* 2005 as cited in Aldenkamp et al 2009).
- The life-limiting nature of the disease and issues relating to service provision also impact psychosocially (Aldenkamp *et al* 2009).

Assessment

Key stages of assessment are usually at diagnosis, prior to school entry and post exacerbation of the condition. Assessments should involve areas of emotional adjustment and coping, neurocognitive functioning (Donders and Taneja 2009), speech and language development,

the potential presence of autism and social support. Routine screening of patient and family is necessary (Bushby *et al* 2010 Part 1).

The Personal Adjustment and Role Skills Scale (PARS-III) measures psychosocial adjustment in children with chronic physical illness (Aldenkamp *et al* 2009). Psychosocial adjustment is the adaptive task of managing upsetting feelings and frustrations secondary to the illness and keeping an emotional balance (Aldenkamp *et al* 2009). The PARS-III has been shown to be a reliable and valid measure for screening psychosocial adjustment in DMD (Aldenkamp *et al* 2009).

Intervention

Interventions used for psychosocial issues in DMD should be the same evidence based interventions that are used in the healthy population. It is crucial to emphasise preventative measures and early treatment in order to improve outcomes. Refer to Busby *et al* (2010) Part 1 for further information.

The TREAT-NMD network of excellence for neuromuscular diseases in Europe has put forward brief standards of care for DMD. Under psychosocial interventions, it is recommended that social (information, advocacy and advice) and psychological support should be provided during times of changing needs and distress (Bushby and Sejerson 2009). Psychological support should be provided during times of emotional/behavioural problems. Learning difficulties should be identified early and advice given to parents (Bushby and Sejerson 2009).

Patient Perspective

In a survey of 65 Danish adults living with DMD, who were aged between 18 -42years, daily life was described positively although the majority longed for a loving relationship and only few have the opportunity to follow further educational or employment opportunities (Rahbek *et al* 2005 as cited in Aniapravan *et al* 2012)

Research by Abbot and Carpenter (2010) focuses on the transition from childhood to adulthood for young men with Duchenne – how it affects their families and the key professionals who support them. It explored transition in two senses:

- firstly the social and psychological aspects of this major life stage and
- secondly the transition from one set of services to another.

This research provides insight into the psychosocial aspect of DMD at an important transitionary stage in their lives. For further details: http://www.bristol.ac.uk/norahfry/research/completed-projects/becominganadult.pdf

Stuart Reid a man living with Duchenne was involved in the above research. Stuart has spoken about how DMD affects him socially, in an interview available: *http://www.muscular-dystrophy.org/about_muscular_dystrophy/yourstories/interviews/3878_a_life_worth_living_stuart_reid*

"The actual condition is rarely what bothers me; it is the way it impairs my relationships with other people. Male relationships are built on doing things together rather than talking so being unable to take part can affect this. The thing I get most upset about is not having any sort of love/sex life like other young men"

Table 3.3.1 DMD Management

	Stage 1: Presymptomatic	**Stage 2: Early Ambulatory**	**Stage 3: Late Ambulatory**	**Stage 4: Early Non-Ambulatory**	**Stage 5: Late Non-Ambulatory**
Clinical Impression	May show symptoms of developmental delay but no gait disturbance	Gower's Manoeuvre Waddling gait Toe-walking Can climb stairs	Increasingly laboured gait Decreased ability to climb stairs and get up from floor	Loss of lower limb function May be able to self-propel wheel chair Able to maintain posture but development of abnormalities (scoliosis)	Upper limb function and postural control limited
Diagnosis	May or may not be diagnosed depending on family history and whether CK test carried out.		Likely to be diagnosed at this stage due to progressive nature of DMD.		
Neuromuscular Management	Anticipatory planning for progression of symptoms Ensure immunization Education and support to parents	On going assessment by MDT to ensure disease progression is as expected and to identify and prevent secondary impairments where possible At least six-monthly assessment of function, strength and range of movement. Determine plateau phase and the need for steroid intervention. On going management of steroid regime and side-effect management.			
Orthopaedic Management	Night time splinting if indicated Surgery rarely indicated		Consideration of surgical options for Achilles tendon contractures in	Monitoring for scoliosis: Intervention with posterior spinal fusion in defined situations.	

	Monitoring	certain situations	Possible intervention for foot position for wheelchair positioning
Respiratory Management	Ensure immunization Baseline respiratory assessment	Monitor respiratory health and function Low risk of impairment.	Closely monitor respiratory function on a regular basis. Increasing risk of impairment
Cardiovascular Management	Baseline cardiac assessment	Annual assessment of cardiovascular function (EMG and echo)	Increased vigilance on assessment due to increased risk of cardiac problems Requires intervention even if asymptomatic. Use of standard heart failure interventions with deterioration of function
GI Management	Ongoing assessment for normal age-related weight gain Dietary management for those over/under weight Speech and Language management if indicated		Strict attention to possible dysphagia
Psychosocial Management	Family support EI to facilitate normal behavioural and learning development	Ongoing assessment and intervention for learning, behaviour and coping Promote independence and social development	Transition planning to adult services

3.4 The Role of Physiotherapy in DMD Management

The physiotherapist is involved in the management of the patient with DMD from the point of diagnosis onwards. Ongoing assessment and monitoring of muscle strength, ROM, tone and symmetry, the patient's posture and his/her overall function is required at least every 4months. Effective planning and setting realistic goals can help facilitate smooth transitions from stage to stage.

3.4.1 Assessment

- Motor Development:
  - ❖ Peabody Developmental Motor Scales-2
    Valid, reliable and standardised, (Provost *et al* 2004)
- Muscle Power, length and tone.
- Functional Ability: Walking; Climbing stairs; Getting up from floor; Transfers.
- Use of mobility aids and assistive appliances
- Balance and Coordination
- Posture
- Joint Rang of Motion

3.4.2 Physiotherapy Interventions

Facilitating gross motor development

With young children who present with gross motor delay, the physiotherapist should demonstrate exercises and facilitation techniques to the caregivers to help the infant/child attain their developmental milestones.

Stretching and orthoses

Passive stretching and the use of night-time orthoses such as AFO's is often indicated during the ambulatory phases of DMD to help delay the development of contractures. Active, active-assisted and/or passive stretching should be done 4-6 times per week minimum, Bushby *et al* (2010). In ambulant children, night-time splints can be worn when there is a loss of ROM, e.g. ankle dorsi-flexion. Daytime AFO's are not recommended before loss of ambulation as they may impede walking ability. In non-ambulant children, sitting AFO's are indicated to help prevent the development of painful contractures.

Maintaining strength and promoting function

Physiotherapy input is essential for the maintenance of muscle function in DMD, Eagle (2002). The physiotherapist should focus on symmetrical muscle strength because weak and underused muscles can lead to the development of asymmetrical contractures predisposing the child to a spinal scoliosis. For further information refer to the exercise section.

Respiratory Care

Chronic respiratory failure is often the cause of mortality in DMD. The physiotherapist plays a key role in respiratory management of DMD. For further information see the respiratory management section above.

Education

Like each member of the MDT, the physiotherapist should explain his/her role in the management of DMD. Throughout the disease process, parents, carers and patient should be thoroughly educated on the disease process, exercise, energy conservation, posture and spinal alignment. To optimise QoL, participation and independence should be encouraged wherever possible.

3.5 Key References

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