

Genetic Disorders

What is a genetic disorder?

Genetic disorders are diseases that are caused by an abnormality in the genome. Abnormalities can range from a small mutation in a single gene to the addition or subtraction of an entire chromosome or set of chromosomes. Most Genetic disorders are present from birth and are quite rare, often affecting only one person in every several thousand or million. Genetic disorders are usually heritable and are passed down from the parents' genes. Other defects are caused by new mutations (changes) to the DNA. In these cases the defect will only be heritable if it occurs in the germ line (i.e. a line of biological cells called germ cells that give rise to gamete - cells that fuse during fertilisation - e.g. ovum and sperm).

What are the common features of genetic disorders?

There are many different types of genetic disorders, all with different features of their own. For specific details on the individual features of specific genetic disorders go to the association websites for each genetic disorder or Wikipedia. Some of the commonly seen genetic disorders include:

- Prader-Willi Syndrome
- Williams Syndrome
- Angelman Syndrome
- Kabuki Syndrome
- Smith Magenis CHARGE

- Cornelia deLange Syndrome
- CRIduCHAT Syndrome
- Microcephali

Common difficulties often (but not always) experienced by the child with a genetic disorder:

- May lack interest/motivation in activities, or be hard to engage in particular activities they find difficult or in which they have experienced failure.
- May have poor social skills.
- Easily frustrated when completing tasks.
- Easily distracted.
- Reduced self-esteem.
- Poor understanding of the conventions of social interaction.
- Poor articulation of sounds.
- Difficulties reading and writing.
- Immature play skills/interests.
- Anxiety when asked to participate in difficult activities.
- Tend to seek out younger children to play with as their skills are of a similar level and they feel more confident playing with them.
- May complain that 'this is too hard' or 'I can't do it' when presented with motor activities.
- May be resistant to changes in how or when tasks are done as changes present new situations/tasks that require planning and new learning.
- Poor non-verbal communication.
- Poor understanding of instructions, questions or jokes.
- Difficulty with fine and gross motor skills.
- Difficulties accessing the school curriculum.

Management strategies that support the child with a genetic disorder (at preschool, school and/or home):

- Provide lots of praise and encouragement.
- Use visual cues to support organisation and planning as well as attention

to task.

- Allow extra time to process and learn when presenting the child with a new task.
- Recognise that additional practice is often required on an on-going basis to recall a previously mastered task.
- Break large tasks into smaller ones (wherever possible) even if it seems silly (not only does this support skill development, but also reduces the heightened anxiety that is common with motor planning challenges).
- Provide opportunities to succeed by simplifying activities.
- Use simple language and instructions.
- Recognise and reinforce the child's strengths.
- Set realistic and achievable goals for all task performance and completion.

Occupational Therapy approaches and activities that can support the child with a genetic disorder and/or their carers include:

- **Expanding abilities:** Developing a gradually broadening range of skill areas.
- **Social stories:** Providing ideas and education around social story development.
- **School transition:** Advocating and professionally supporting the transition to school and liaising with teachers, as required.
- **Visual cues** can be used to support routine and to introduce new activities, or a change in tasks.
- **Gross and fine motor skills:** Determining the current age level of a child's gross and fine motor abilities.
- **Devising goals:** Setting functional and achievable goals in collaboration with the child, parents and teachers so that therapy has a common focus beneficial to everyone involved.
- **Educating** parents, carers, teachers and others involved in the child's care about the child's Genetic Disorder and the age appropriate skills a child should be demonstrating.
- **Direct skill teaching** through a task based approach.

- **Management strategies:** Providing management strategies/ideas to assist the child in the home, at school and in the community.
- **Task engagement:** Providing alternative ways to encourage task engagement.
- **Developing underlying skills** necessary to support whole body (gross motor) and hand dexterity (fine motor) skills, such as providing activities to support:
 - balance and coordination
 - strength and endurance
 - attention and alertness
 - body awareness
 - movement planning

Speech Therapy approaches and activities that can support the child with a genetic disorder and/or their carers include:

- **Speech and language assessment** to help the family to understand how the child is processing, understanding, learning and using language and communication.
- **Communication strategies:** Providing the family with strategies and techniques to increase and enhance communication with the child.
- **Daily activities:** Helping the child to understand the environment, routines and language.
- **Developing language:** Helping the child to understand and use richer language and to use language more spontaneously.
- **Conversation skills:** Developing conversation skills (e.g. back and forth exchange, turn taking).
- **Concept skills:** Developing concept skills, especially abstract concepts, such as time (e.g. yesterday, before, after).
- **Visuals** can be used to help with understanding and the child's ability to express their needs, wants, thoughts and ideas.
- **Social skills:** Development of social skills (i.e. knowing when, how to use language in social situations).
- **Enhancing verbal and non-verbal communication** including natural

gestures, speech, signs, pictures and written words.

- **Visual strategies:** Using visual information to help understand, organise and plan the routine for the day.
- **Liaising with educational staff** regarding the nature of the difficulties and ways to help the child to access the curriculum.

Why should I seek therapy if my child has a genetic disorder?

Diagnosis alone is NOT the solution. It simply opens the door to getting the help that is needed by arming all involved with the relevant information.

The 'help' still needs to be provided. The help that is provided (at least from a therapy perspective) will reflect:

- First and foremost what medical intervention is needed.
- What the parents/teachers/carers biggest concerns are for the child (i.e. what are the most significant functional challenges).
- The specific areas that are problematic to the child (which will vary even within children with the same diagnosis).
- The capacity of the child's environments to meet the child's needs.

If left untreated the child with a genetic disorder may have difficulties with:

- Following instructions within the home, kindergarten or school environment.
- Vocabulary whereby a child cannot clearly get their message across due to limited word knowledge.
- Understanding jokes and figurative language during interactions with others, and when watching TV shows and movies and reading books.
- Learning to talk, speech intelligibility and clarity.
- Managing a full school day due to poor strength and endurance.
- Participating in sporting activities leading to an inactive lifestyle, increasing the risks of other health related issues such as obesity,

diabetes, cardiovascular disease or similar conditions.

- Self esteem and confidence when they realise their skills do not match their peers.
- Bullying when others become more aware of the child's difficulties.
- Fine motor skills (e.g. writing, drawing and cutting) due to poor core stability, meaning they do not have a strong base to support the use of their arms and hands.
- Completing self-care tasks (e.g. doing up shoelaces, buttons, zips, using cutlery).
- Self regulation and behaviour as the child is unable to regulate themselves appropriately to settle and attend to a task for extended periods of time.
- Accessing the curriculum because they are unable to attend to tasks long enough to complete assessment criteria.
- Sleep habits, impacting upon skill development due to fatigue.
- Social isolation because they are unable to cope in group situations or busy environments, impacting on their ability to form and maintain friendships.
- Anxiety and stress in a variety of situations leading to difficulty reaching their academic potential.
- Social communication, such as eye contact, appropriate distance when talking to someone, turn-taking within a conversation.
- Academic performance: Developing literacy skills such as reading and writing and coping in the academic environment.
- Academic assessment: Completing tests, exams and academic tasks in higher education.

More specific implications of not seeking treatment will be influenced by the common difficulties that are most influencing your individual child.

For more information see the relevant fact sheets under areas of concern or refer to the other relevant resources section below.

What does this diagnosis of a genetic disorder really mean

for the child?

Diagnoses are used to label a specific set of symptoms that are being experienced by a child. This label then helps to narrow down and specifically tailor what:

- Other issues commonly occur simultaneously.
- Medication might be appropriate.
- Therapies might help the child (e.g. Medical, Occupational Therapy, Speech Therapy, Psychology).
- Course of intervention (medical and/or allied health) might be and what outcome might be expected (prognosis).
- Can be done to help the child.

A diagnosis helps the child and their carers (parents, teachers, health professionals, carers) to:

- Access information about the relevant cluster of symptoms.
- Communicate the salient features of the child's challenges to all people involved in the child's care.
- Possibly interpret certain behaviours differently in light of the diagnosis.
- Obtain information about what can be done to help the child.
- Determine specifically where and how to help the child.
- Access funding or services that might not otherwise be accessible.

Contact us today to make an initial enquiry or book an assessment for your child
on 1800 KID SENSE (1800 543 736)

