COMPLEX LEARNING DIFFICULTIES AND DISABILITIES RESEARCH PROJECT (CLDD)

RARE CHROMOSOME DISORDERS

What are rare chromosome disorders?

The term, 'rare chromosome disorders', refers to conditions which:

1. occur due to missing, duplicated or re-arranged chromosome material
2. have a low prevalence rate (thus not including chromosomal disorders such as Down syndrome).

Chromosomes are structures found in the nuclei of cells in human bodies. Each chromosome contains thousands of genes which determine how we grow and develop. A typically developing person will have 23 pairs of chromosomes with one member of each pair being inherited from each parent, giving a total of 46 individual chromosomes. Two of these are the sex chromosomes, which determine whether we are female (XX) or male (XY). The remaining 44 chromosomes are grouped in 22 pairs numbered 1 to 22. The arms of a chromosome are called 'p' (shorter arm) and 'q' (longer arm) (see Figure 1).

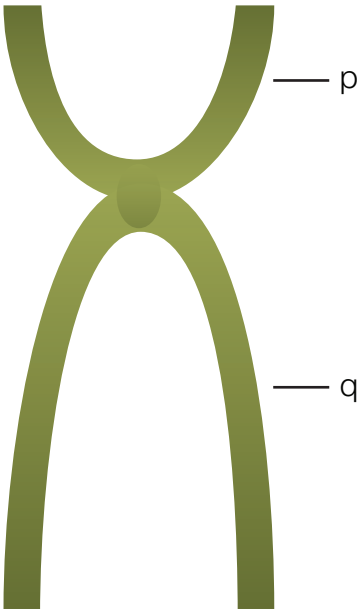

Figure 1. Diagram of a chromosome

Even when people are identified as having a similar condition, the way in which it affects each person may still vary a great deal. If enough children are born with the same chromosome disorder, and present a similar pattern of characteristics, it may be called a syndrome. Rare chromosome disorders account for at least one in every 200 live births, with babies either having symptoms of the disorder from birth or early childhood, or being carriers of a chromosomal abnormality and experiencing the effects when they try to reproduce in later life.

'Sex chromosome disorders' is an umbrella term for disorders where there are too many sex chromosomes. Each disorder is gender-specific, and the prevalence rate is one in 1,000 live births. Students with these disorders generally show only subtle physical features and most achieve within age-related norms at school, perhaps with mild learning difficulties.

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Possible indicators of rare chromosome disorders

Rearranged abnormality: Students with a rearranged abnormality may not experience any symptoms, but might have problems in reproduction. Some people consider genetic counselling when they are planning a family to assess the potential impact their chromosomal abnormality may have on their children..

Missing or duplicated chromosome material: For students with missing or duplicated chromosome material, the effects will vary, but symptoms could include physical and/or health problems, learning disability and maybe challenging behaviour. The combination and severity of symptoms will vary depending on which sections of chromosomes are involved, and the ways in which they are different.

Chromosome loss or gain: Students with a loss or gain of chromosomes will experience some degree of learning disability and developmental delay. This is thought to be more serious than the presence of an extra copy of the same part.of an extra copy of the same part.

Implications for teaching and learning

The implications for teaching and learning for these students varies from one disorder to another, and not every student with the same disorder will present in the same way.

Strengths across the range of disorders may include:

- conveying emotion through facial expression, vocal noises, gestures and body movements imitation
- good memory, particularly for faces and places
- enjoyment of books and music
- happy, sociable and enthusiastic disposition when young
- maths calculations, rote memory, spelling and written language, decoding words and basic reading (particularly in students with DiGeorge Syndrome).

The table below outlines the main **areas of difficulty** relating to some examples of rare chromosome disorders. (As you will see, some of the syndromes do not have names as such, but are referred to by the chromosome number that causes the abnormality.)

Type of difficulty	Examples of rare chromosome disorder(s) in which the difficulty may be present
Seizures	1p36; Idic15
Hypotonia or flopiness	1p36; 22q13 deletion; Idic15; Jacobsen; DiGeorge
Difficulties with feeding	1p36
Heart problems	1p36; DiGeorge

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Hearing and/or visual impairments	1p36
Paris-Trosseau syndrome (bleeding disorder causing bruising and heavy blood loss)	Jacobsen
Distinctive facial features	1p36, Cri du chat, 22q13 deletion, Idic15, Jacobsen, DiGeorge
Microcephaly (unusually small head)	Cri du chat
Low birth weight/small stature/poor immunity	Cri du chat, Jacobsen, DiGeorge
Developmental delay	1p36, 22q13 deletion, Idic15, DiGeorge
Learning disability	1p36, Jacobsen, DiGeorge
Sensory processing difficulties, with symptoms which include tactile defensiveness, chewing non-food items and teeth grinding	1p36, 22q13 deletion, Idic15
Social interaction	1p36, 22q13 deletion, Idic15, DiGeorge
High pitched 'cat-like' cry	Cri du chat
Speech and language delays	Cri du chat, 22q13 deletion, Idic15, Jacobsen, DiGeorge
Hyperactivity/inattentive	Cri du chat, Idic15, Jacobsen, DiGeorge
Poor concept of danger	Cri du chat

Supporting these students

Approaches may vary according to the particular disorder and the individual student. (A guide for each disorder listed above can be found on the information sheet relating to this topic.) However, the following approaches will be effective for many students:

1. The appropriate use of music, light and tactile resources, with an emphasis on visual learning is thought to assist learning and the control of seizures (take specialist advice in use of light).

2. Patience, repetition and lots of encouragement in a calm, structured learning environment.

3. Concrete resources, including computer based learning, to overcome difficulties with abstract concepts, such as time, money, shape, colour and size. Direct instruction rather than reliance on discovery may work well. However, creative projects have been found to be stimulating.

4. Speech and language therapy to aid communication and reduce frustration, particularly as some students will have a cleft palate and feeding difficulties. Signing may be effective, but some students may have poor motor control which will make this difficult.

5. Computer touch screens, voiced based systems and picture exchange systems (such as PECS)

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to increase communication skills.

6. Adaptive sports, music therapy and sensory integration support (from an occupational therapist) to increase awareness and desire to communicate.

7. Weighted blankets, hiking and wearing backpacks, deep pressure massage and rolling games to stimulate balance and body awareness (an occupational therapist should advise on particular techniques for individual students).

8. Share interventions and strategies used in school with families and other professionals involved to ensure consistency and understanding.

When considering any of these support strategies, bearing in mind the student's individual needs, preferences and interests is essential to increasing engagement. The student themselves, families and staff that know the student well can all help to inform on what these may be.

Key references

A full reference list can be found on the Information Sheet relating to this area.

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Information and support for specific disorders can be found online at www.rarecromo.org