

COMPLEX LEARNING DIFFICULTIES AND DISABILITIES RESEARCH PROJECT (CLDD)

RARE CHROMOSOME DISORDERS

What are rare chromosome disorders?

Chromosome disorders are ones that occur when one or more chromosomes do not develop normally.

Rare chromosome disorders are ones where there is a very low incidence rate. So more usual disorders caused in this way, such as Down syndrome, are not included.

Strengths

These students may have strengths in some of the following areas:

- 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 students with DiGeorge syndrome).

Difficulties

These are set out in the table below. (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 floppiness	1p36; 22q13 deletion; Idic15; Jacobsen; DiGeorge
Difficulties with feeding	1p36
Heart problems	1p36; DiGeorge
Hearing and/or visual impairments	1p36
Paris-Trousseau 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

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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 learners

The following approaches may be helpful for students with rare chromosome disorders, although much will depend on which disorder they have, how it presents in each individual and their interests and preferences:

- 1 A calm, structured learning environment. These students are likely to need patience, repetition and lots of encouragement.
- 2 Speech and language therapy will help them to communicate and reduce frustration, particularly for those who have a cleft palate and feeding difficulties. Signing may help, but students with poor motor control may not be able to manage to sign themselves.
- 3 Computer touch screens, voice based systems and picture exchange systems (such as PECS – Picture Exchange Communication System) can be used to improve communication skills.
- 4 Involvement in sports, music therapy and sensory integration support (which may be available from an occupational therapist) will all increase the desire to communicate.
- 5 An emphasis on visual learning, with resources they can handle, will help students understand what is being taught. The appropriate use of music and light may help to control seizures, but take specialist advice on the use of light.
- 6 Computer based learning can help to overcome difficulties with abstract concepts such as understanding time, money, shape, colour and size. Creative projects can work well
- 7 Occupational therapists and physios may also be able to advise on using weighted blankets, wearing backpacks, deep pressure massage and rolling games, which can be used to help balance and develop body awareness. These should not be used without appropriate advice.