Genetic and Neurodevelopmental Conditions Linked to IDD

Angelman Syndrome

Angelman syndrome is a genetic disorder that occurs because of a lack of expression of a gene on a maternally inherited chromosome. Angelman syndrome is associated with severe intellectual disability and characteristic facial features.

People with the condition typically have physical challenges, such as delayed motor milestones, movement or balance disorders, limited or no speech, seizures, and sleep disturbances. Common behavioural characteristics associated with the condition include apparent bouts of excessive, often inappropriate laughter, easily excitable, and repetitive or stereotyped behaviours (such as hand flapping and mouthing).

Autism Spectrum Disorder

Autism spectrum disorder (ASD) is a condition that typically appears early in childhood development. The term “spectrum” refers to the wide range of symptoms and severity including impairments in social interaction and communication, and restricted, repetitive behaviours or interests. People with ASD can have different levels of difficulties in these areas. These difficulties can interfere with their ability to function in social, academic, and employment settings.

People with ASD are also more likely to have psychiatric problems such as anxiety, depression, obsessive-compulsive disorder, and eating disorders. People with ASD can have difficulties in understanding and using non-verbal social cues such as eye contact, facial expressions, gestures, and body language.

Cerebral Palsy

Cerebral palsy is an umbrella term, which means it refers to a group of disorders and symptoms. While all the possible symptoms, disabilities, and complications are related, one person’s experience is often very different from another’s.

Cerebral palsy is the most common disability that impacts movement and motor skills. It is a neurological disorder that affects motor skills, movements, and muscle tone. Brain damage is the underlying cause. The damage may occur while the baby is still in utero, during labor and delivery, or shortly after birth.

Having cerebral palsy can lead to a number of other medical conditions, depending on the severity of the disorder, such as speech problems, learning disabilities, cognitive impairments, problems with hearing and vision, epilepsy, emotional and behavioural issues, spinal deformities, and joint problems.

Down Syndrome

Down syndrome (or Trisomy 21) is a genetic condition that is associated with intellectual disability. The condition is caused by being born with an extra part or full chromosome. People with Down syndrome have a characteristic facial appearance.

People with Down syndrome may have a variety of physical health issues. About half of all affected children are born with a heart defect. Digestive abnormalities, such as a blockage of the intestine, also occur sometimes, but these are not as frequent. Adults with Down syndrome can have thyroid problems, sleep apnea, and may also develop Alzheimer’s disease as they age.

Fetal Alcohol Spectrum Disorder

Fetal Alcohol Spectrum Disorder (FASD) is a term used to describe the range of effects that can occur in an individual whose mother consumed alcohol during pregnancy.

When a woman drinks alcohol while pregnant, her fetus is exposed directly to alcohol through her bloodstream. Alcohol can interfere with the growth and development of all fetal body systems. The developing central nervous system (the brain and spinal cord) is most vulnerable to the damaging effects of alcohol. These effects, which can vary from mild to severe, may include physical, mental, behavioural, and/or learning disabilities with possible lifelong implications.

Fragile X Syndrome

Fragile X syndrome occurs in individuals with a specific genetic mutation and is the most common type of hereditary intellectual disability. Typically, males with this condition have moderate intellectual disability and females with the condition tend to have mild intellectual disability. Some males with this condition will have a large head, long face, prominent forehead and chin, protruding ears, joint laxity, and large testes after puberty. Behavioural abnormalities, including autism spectrum disorder, are common in people with this condition as well.

Prader-Willi Syndrome

Prader-Willi syndrome is associated with weak muscle tone (hypotonia) and feeding difficulties in early infancy. After infancy, individuals develop excessive eating patterns and usually develop obesity, unless their eating is controlled by others. Short stature is common (if not treated with growth hormone); characteristic facial features, strabismus (a vision problem), and scoliosis are often present.

People with Prader-Willi syndrome typically have delays in their motor and language skills. Everyone with this condition experiences some degree of cognitive impairment. Behavioural problems such as temper tantrums, stubbornness, and obsessive-compulsive behaviour are often present as well. Hypogonadism (the diminished functioning of the testes or ovaries), is present in both males and females and manifests as genital hypoplasia (the underdevelopment or incomplete development of a tissue or organ), incomplete pubertal development, and infertility in most.

References:

https://www.ncbi.nlm.nih.gov/gtr/conditions/C1510586/
https://ghr.nlm.nih.gov/condition/down-syndrome
http://ddprimarycare.surreyplace.ca/tools-2/health-watch-tables/
https://www.cerebralpalsyguidance.com/cerebral-palsy/