Genetics and Paediatric Health: Section 2

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À l'écoute de notre santé

2.1.1 Chromosonal Conditions

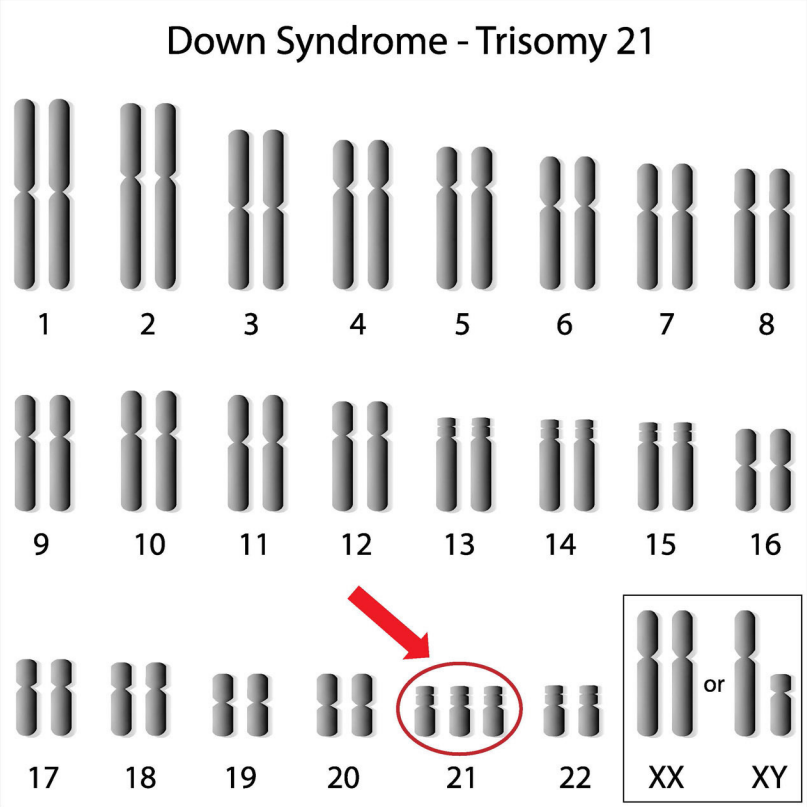

Image credit: Shutterstock

Chromosomal conditions or anomalies occur when there is an abnormal amount of chromosomal material. The usual chromosome complement in each cell is 46. There are 22 pairs of autosomes numbered 1 to 22, with each pair having the same genes. The 23rd pair is called the sex chromosomes, with females having two "X" chromosomes and a male having an "X" and a "Y" chromosome. The "X" and "Y" chromosomes, unlike autosomes, have different genes. There is sometimes an abnormal total number of chromosomes – sometimes an extra chromosome. For example, a child with Down syndrome has three copies of chromosome 21, rather than the usual two, so the condition is called "trisomy 21."

You can find more information about these conditions and how they happen at:

- SickKids, the Genetics section (If a user profile box pops up, you can press 'cancel' or create a user profile to access this site.)
- Centers for Disease Control and Prevention, Facts on Pediatric Genetics

2.1.2 Single Gene (Monogenic) Conditions

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Single gene conditions (also called monogenic conditions) are caused by abnormalities, or mutations, of a single gene. These conditions can be severe or harmless, and though most are rare, some can be common. Familial hypercholesterolemia, a genetic condition that can lead to high levels of a harmful form of cholesterol, is an example of a common single gene condition.

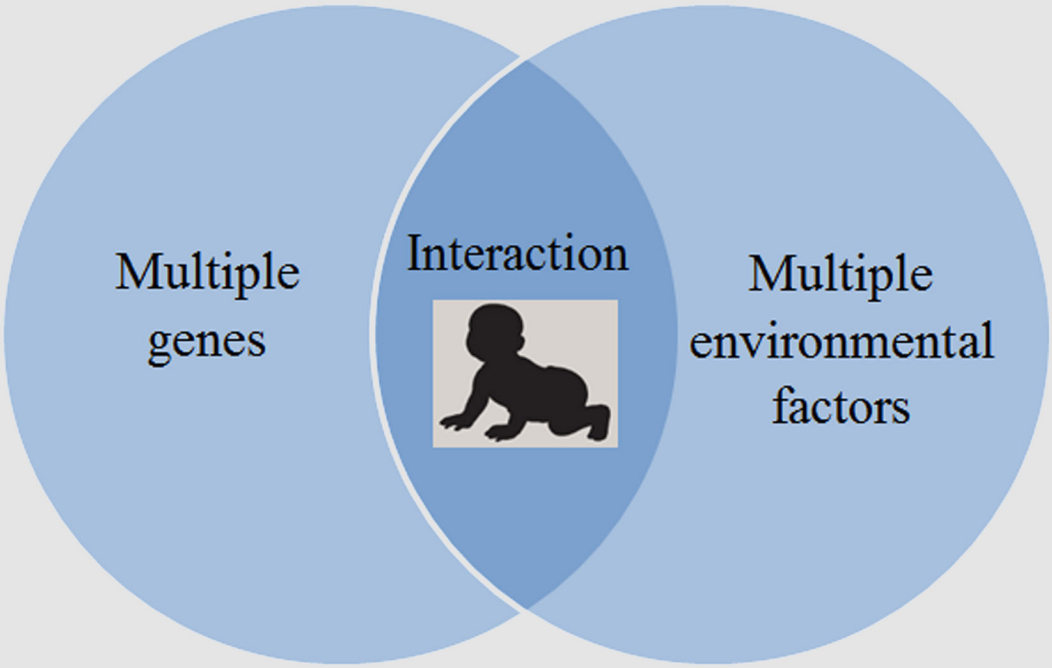
Single gene conditions can have either a dominant or a recessive inheritance pattern. Familial polyposis is a single gene condition that results from a dominant mutation. Dominant inheritance means that the condition occurs when only one copy of a gene pair has a mutation. Familial polyposis often results in cancer of the large intestine (colon) and rectum.

Other single gene conditions only develop when the person inherits two mutations in the same gene on one of the autosomes – one from each parent. These mutations are known as autosomal recessive. An individual with only one copy of a recessive gene mutation does not develop the disease and is instead called a carrier. Some examples of autosomal recessive, single gene conditions are cystic fibrosis, Tay-Sachs disease, and sickle cell anaemia. In these conditions two copies of a mutation in the gene – one from each parent – must be present for a child to develop the condition.

When a mutation exists in a gene on the X chromosome, an X-linked condition may result. An example of an X-linked condition is hemophilia.

For more information about these conditions and how they happen go to: Utah Genetics Education, National Coalition of Professional Education in Genetics, and Genetic Alliance UK.

2.1.3 Multifactorial Disorders or Complex Traits



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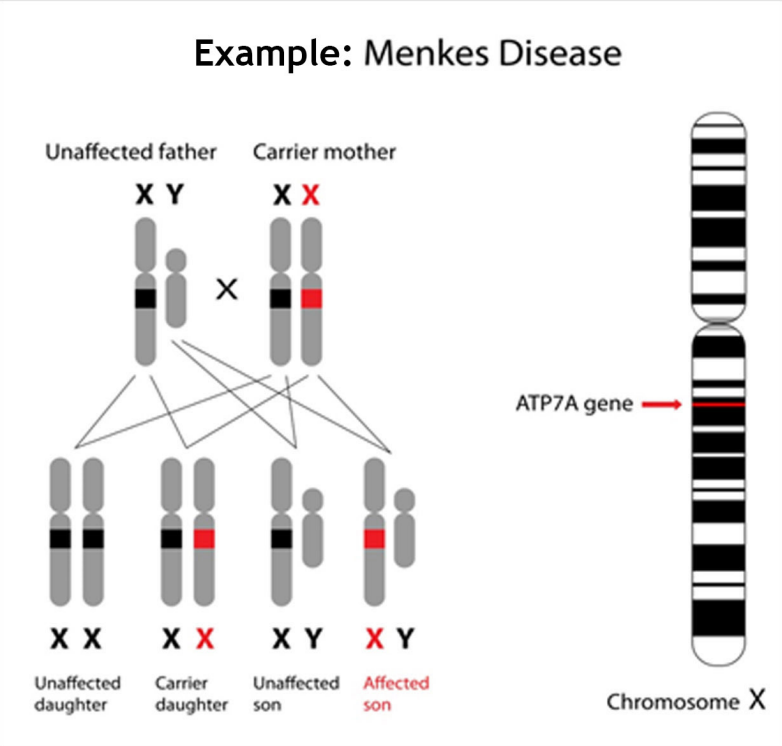
Multifactorial genetic disorders, or complex traits, result from several genes in combination with lifestyle and environmental influences. Multifactorial disorders include diseases like diabetes, many cancers, heart disease, and asthma. Multifactorial disorders account for the largest group of genetic conditions, both in numbers and the impact that they have on child health and the health care system as a whole.

For more information about these conditions and how they happen go to:

- Centers for Disease Control and Prevention, Facts on Pediatric Genetics
- National Coalition of Professional Education in Genetics
- University of Kansas, Genetics Education Center
- Centre for Genetics Education

2.1.4 What Causes Genetic Conditions?



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Menkes disease is a recessive genetic condition caused by a mutation of the gene on the X chromosome that is responsible for the metabolism of copper in the body. Copper levels become abnormally low in the liver and brain and abnormally high in the kidney and intestinal lining. Menkes disease leads to premature birth, floppy muscle tone, developmental delay, seizures, and failure to gain weight and to grow at the usual rate.[1,2,3]

Gene mutations can be inherited from parents; however, mutations in genes and chromosomes can happen unexpectedly and usually for unexplained reasons. Mutations that exist in the parents' genes are inherited mutations. New mutations, which occur during the transmission of genetic material to the next generation, are called de novo mutations. It is important to obtain a complete family history to better understand genetic risk factors for children and their families.

For more information about inheritance see:

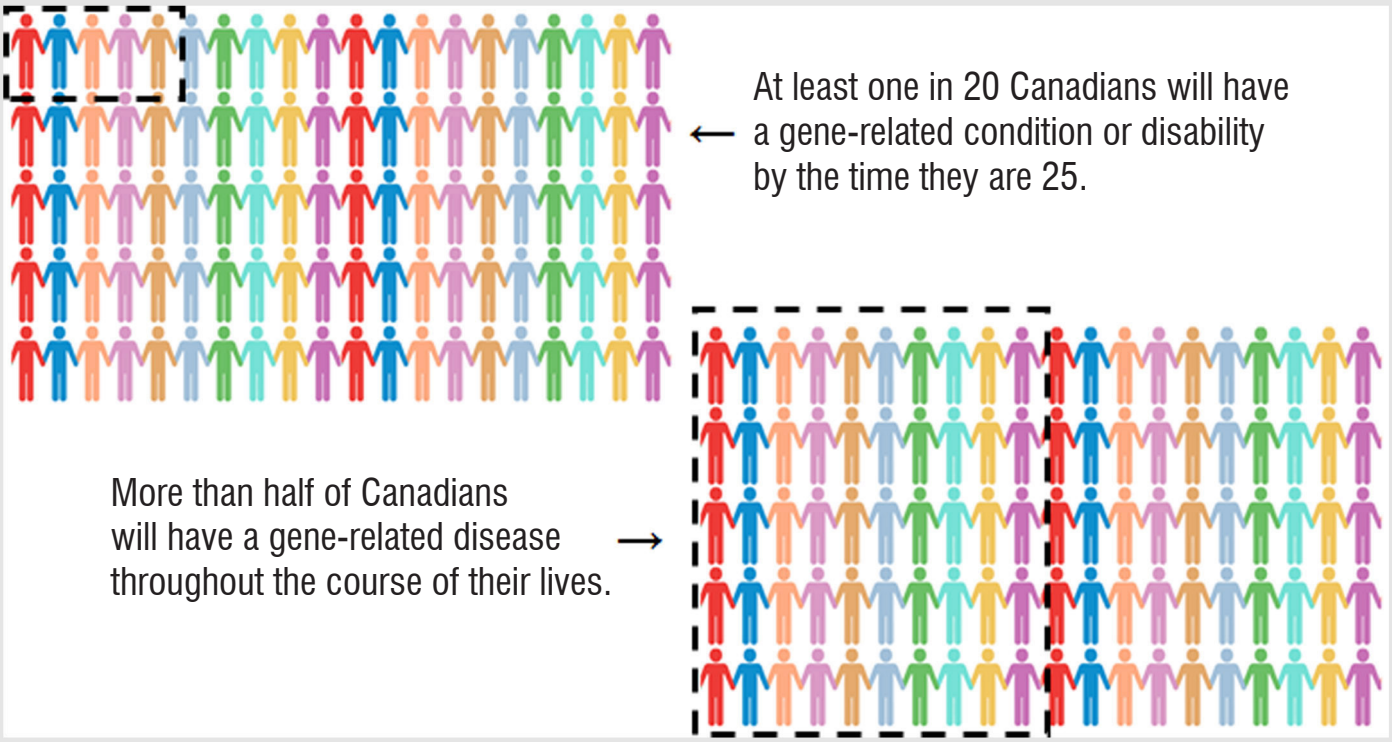
- SickKids, 'Genetics' section (If a user profile box pops up, you can press 'cancel' or create a user profile to access this site)
- Centers for Disease Control and Prevention, 'Pediatric Genetics' section

1 http://ghr.nlm.nih.gov/condition/menkes-syndrome;

2 http://www.merriam-webster.com/medical/menkes'%20disease;

3 http://www.ninds.nih.gov/disorders/menkes/menkes.htm

2.1.5 How Many Children Have Genetic Conditions?



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In Canada, it is estimated that at least one in 20 individuals will experience a gene-related condition or disability by the age of 25. In addition, more than half the population will experience a gene-related disease during their lifetime.[1]

[1] Science Council of Canada. Genetics in Canadian Health Care. Report 42. Ottawa: Science Council of Canada; 1991

2.1.6 What Does this Mean for Children and Families?

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When a genetic diagnosis is suspected or confirmed, the child and his or her family members often require complex support and care, perhaps for a significant period of time. Parents will have questions about what the condition will mean for their child and also how the condition originated, either through genetic inheritance or through a spontaneous genetic mutation, what the risks are of it happening again or in future generations, and what their reproductive choices might be for another pregnancy. They may be concerned about their health and the health of their child, privacy, stigma, or discrimination, and parents often experience guilt.[1, 2] Often other relatives are concerned about themselves or their children. Because there is no cure or effective treatment for many genetic conditions, the disclosure of a genetic diagnosis can be incredibly difficult for all family members involved.

Personal, family, cultural, and religious beliefs influence how families interpret and share information about genetic conditions. There has been an explosion of knowledge and information regarding the genetic bases of disease. It is therefore critical to ensure appropriate clinical services are available to families who need them.

[1] Wilcken B. Ethical issues in genetics. Journal of Paediatrics and Child Health.2011;47:668–71.

[2] Canadian Association of Genetic Counsellors, https://cagc-accg.ca/content/view/12/26/