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.