Human Hereditary Diseases, their Expression and Prediction

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A **genetic disorder** is caused by abnormalities in genes or chromosomes. Some of them, such as cancer, are due to impairments of genetic material acquired in a few cells during life. However, the term "genetic disease" most commonly refers to diseases present in all cells of the body and present since conception, which means that defective genes are inherited from the parents. In this case, the genetic disorder is known as a **hereditary disease**. This can happen unexpectedly when two healthy carriers of a defective recessive gene reproduce, but can also happen when the defective gene is dominant. Most of the 4,000 genetic disorders as known at present, are quite rare and affect one person in every several thousands or millions. However, a number of them, such as familial hyperlipidaemia or hypercholesterolaemia, cystic fibrosis, polycystic kidney disease and Huntington's disease are common enough to raise estimation that 2–3% of births result in babies with either congenital or genetically-determined abnormalities. The number of genetically affected people is even much higher if we include all conditions in which genetic material plays some role. For these reasons medical molecular diagnosis together with genetic counseling plays important role in prediction, elimination and/or in developing strategies for treatment of genetic disorders. Genotyping predispositions for genetically based conditions is a new field of the current medicine which opens ethical questions and concerns for future development.