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Introduction to Genes and Disease

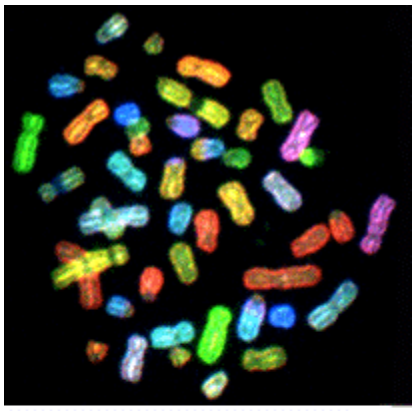
Genes and Disease is a collection of articles that discuss genes and the diseases that they cause. These genetic disorders are organized by the parts of the body that they affect. As some diseases affect various body systems, they appear in more than one chapter.

With each genetic disorder, the underlying mutation(s) is discussed, along with clinical features and links to key websites. You can browse through the articles online, and you can also download a printable file (PDF) of each chapter.

From *Genes and Disease* you can delve into many online related resources with free and full access. For example, you can visit the human genome to see the location of the genes implicated in each disorder. You can also find related gene sequences in different organisms. And for the very latest information, you can search for complete research articles, and look in other books in the NCBI Bookshelf.

Currently over 80 genetic disorders have been summarized, and the content of *Genes and Disease* is continually growing. Your ideas and suggestions are welcome. You can contact us at: info@ncbi.nlm.nih.gov.

Preface



This photograph shows a complete set of chromosomes from an acute promyelocytic leukemia (APL) patient. A new technique called chromosome painting allows visual distinction between chromsomes and can be used to show the chromosome translocations that frequently occur in human cancers. In the case of APL, chromosome 13 is lost, there is a translocation between chromosomes 7 and 15, translocation between chromosomes 11, 15, 17, and between chromosomes 9 and 18. (Look for chromosomes painted with more than one color.) With thanks to Thomas Ried, National Human Genome Research Institute, NIH, for supplying the picture.

The sequence of the human genome is providing us with the first holistic view of our genetic heritage. While not yet complete, continued refinement of the data bring us ever closer to a complete human genome reference sequence. This will be a fundamental resource in future biomedical research.

The 46 human chromosomes (22 pairs of autosomal chromosomes and 2 sex chromosomes) between them house almost 3 billion base pairs of DNA that contains about 30,000 - 40,000 protein-coding genes. The coding regions make up less than 5% of the genome (the function of the remaining DNA is not clear) and some chromosomes have a higher density of genes than others.

Most of the genetic disorders featured on this web site are the direct result of a mutation in one gene. However, one of the most difficult problems ahead is to find out how genes contribute to diseases that have a complex pattern of inheritance, such as in the cases of diabetes, asthma, cancer and mental illness. In all these cases, no one gene has the yes/no power to say whether a person has a disease or not. It is likely that more than one mutation is required before the disease is manifest, and a number of genes may each make a subtle contribution to a person's susceptibility to a disease; genes may also affect how a person reacts to environmental factors. Unraveling these networks of events will undoubtedly be a challenge for some time to come, and will be amply assisted by the availability of the sequence of the human genome.