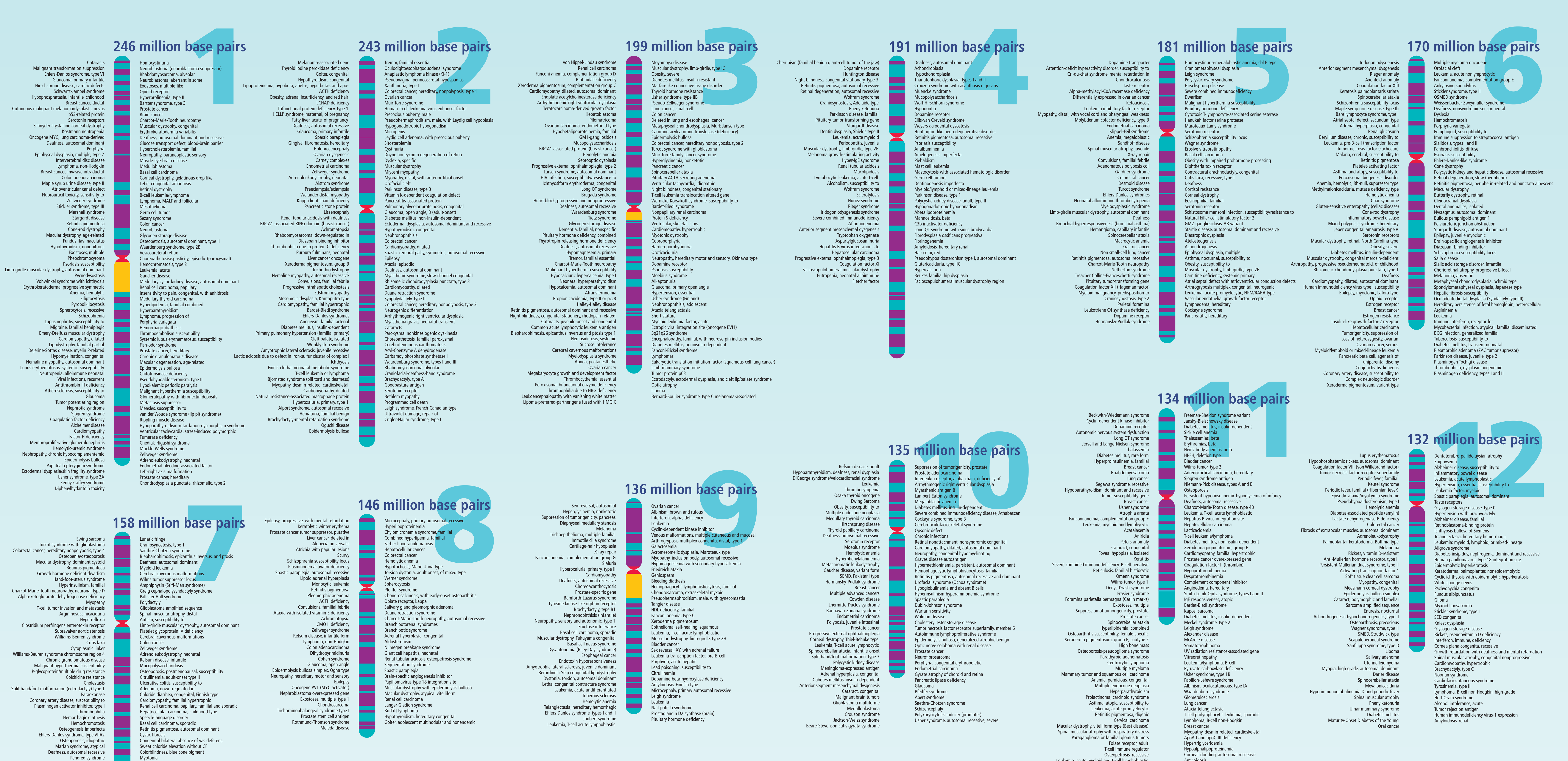
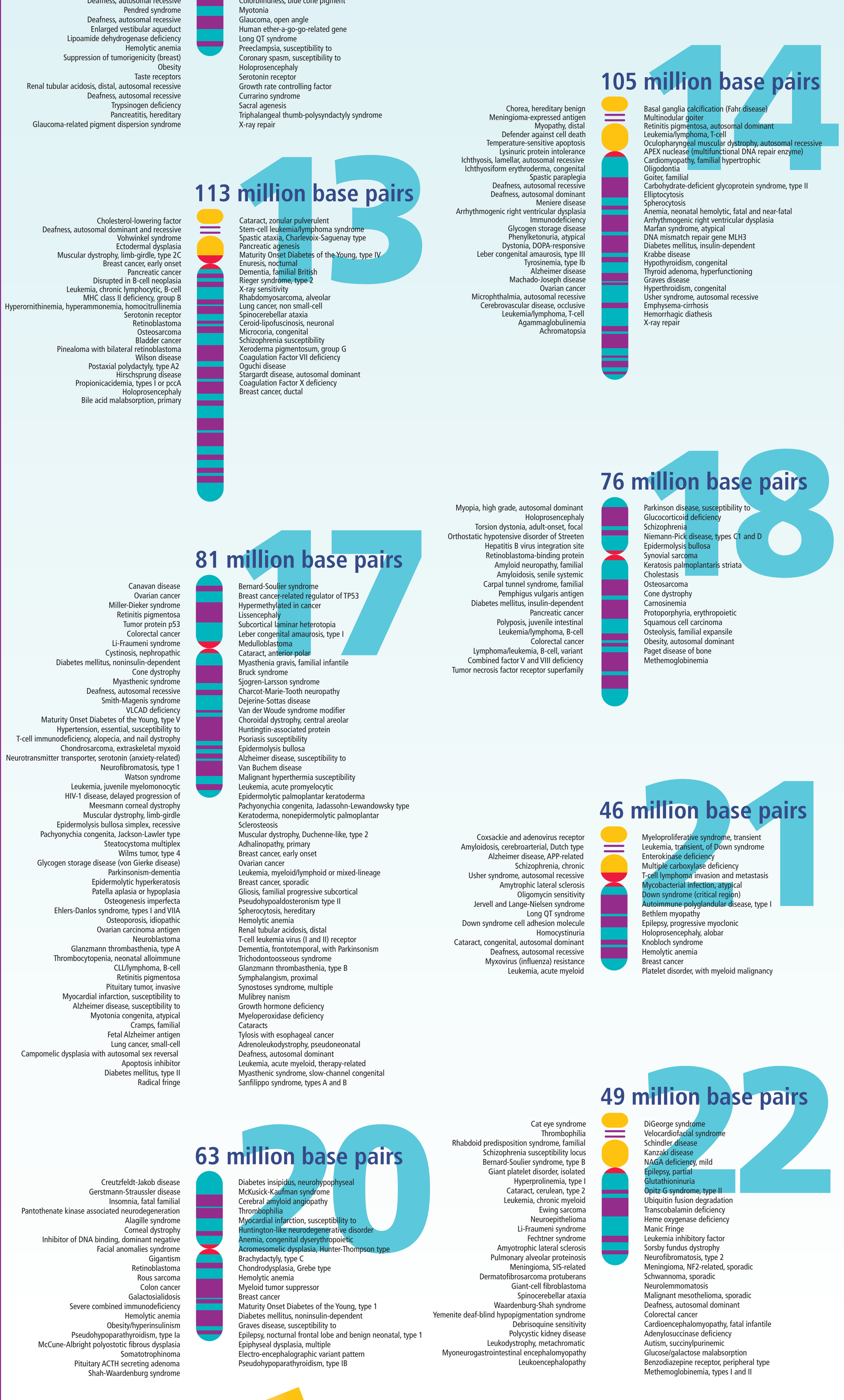
Human Genome Landmarks

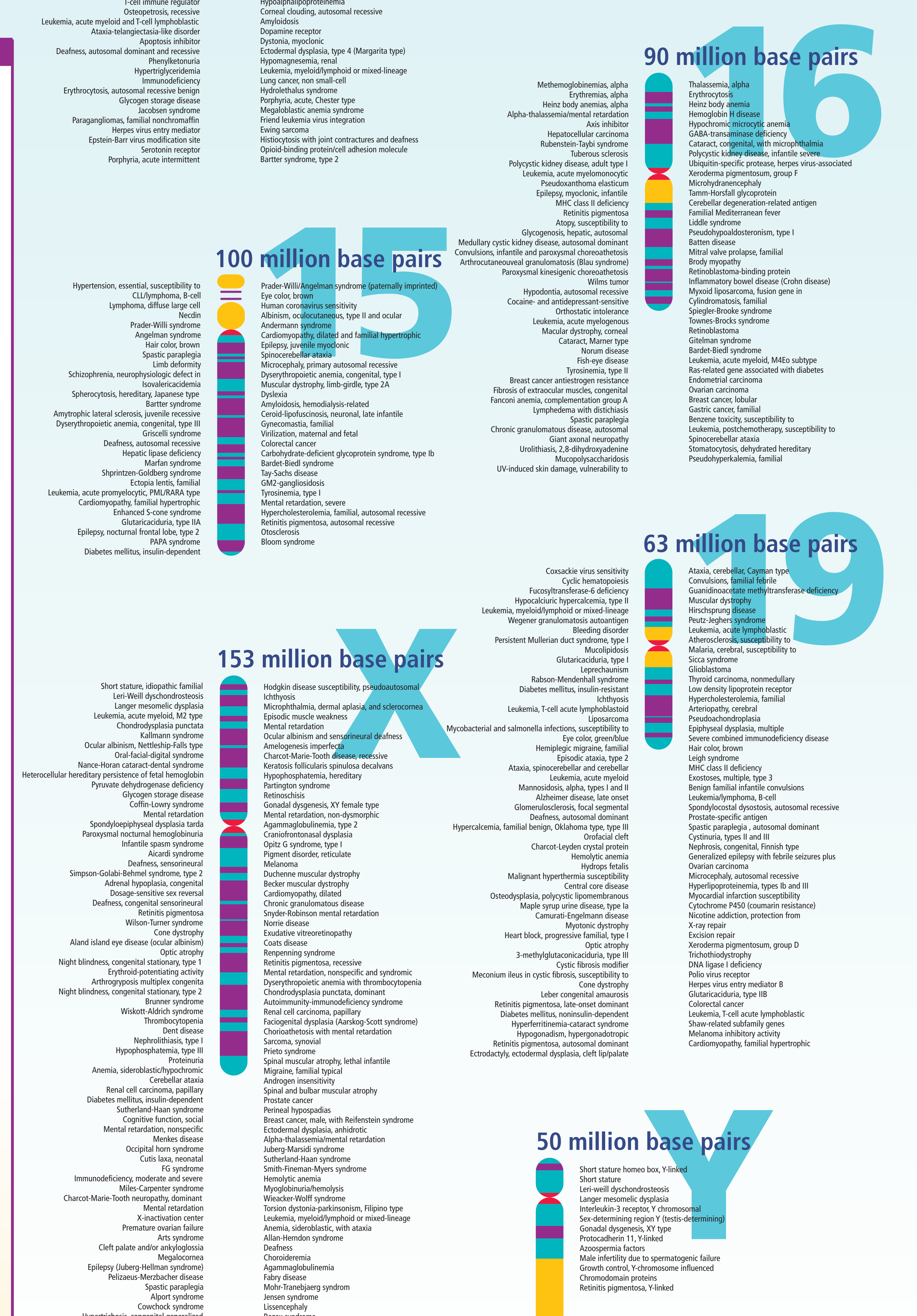
Selected Genes, Traits, and Disorders

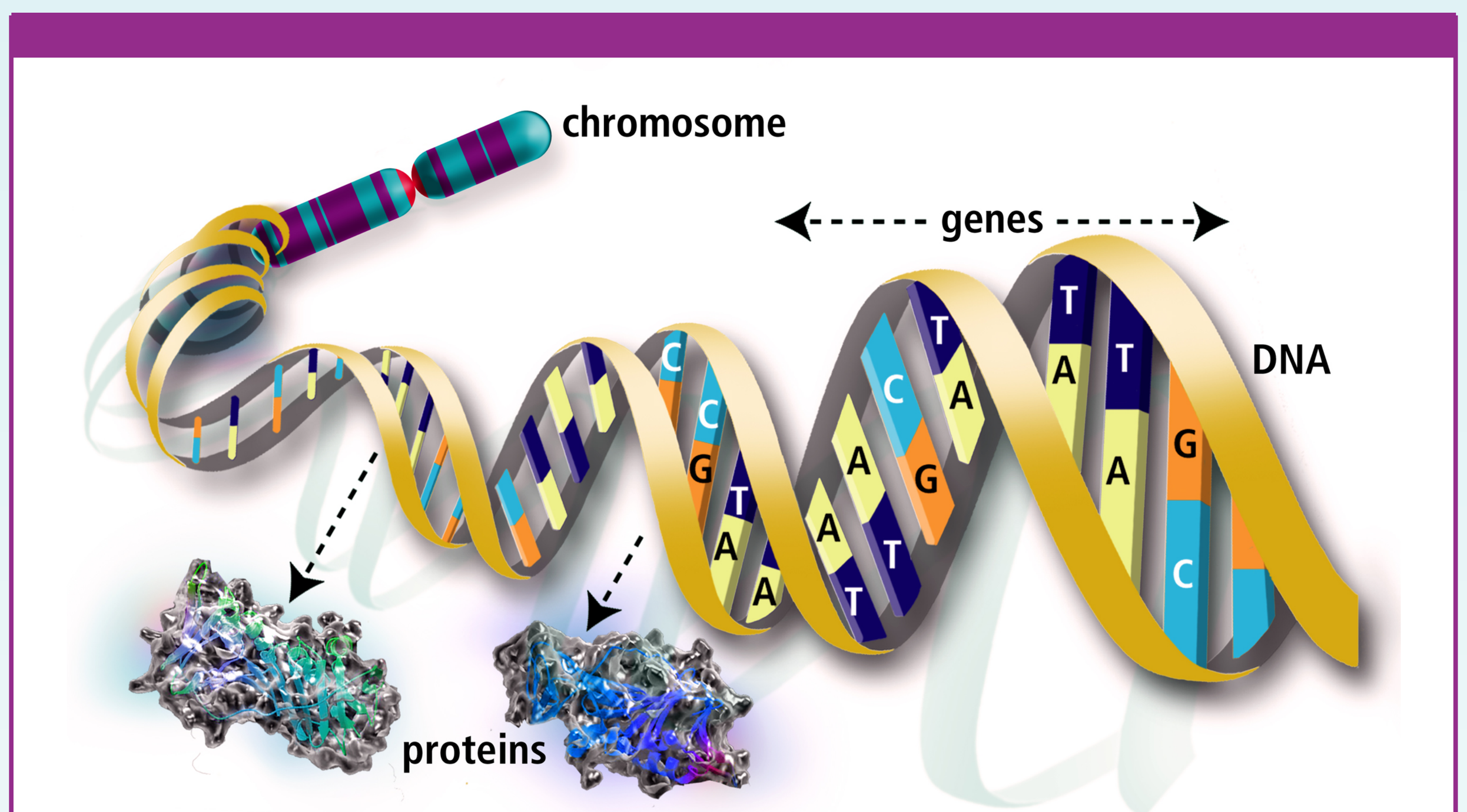
www.ornl.gov/hgmis/posters/chromosome

genomics.energy.gov









A Closer Look

Chromosomes are tightly coiled microscopic structures made up mainly of DNA, which consists of four different building blocks called bases (A, T, C, and G). The four bases are repeated millions of times to form each chromosome. The entire human genome consists of 23 pairs of chromosomes with one chromosome in every pair coming from each parent. With few exceptions (e.g., red blood cells), each of the trillions of cells in the human body contains a complete set of chromosomes—the genome.

The color-banded figures in this poster are simplified representations (ideograms) of chromosomes, which can be distinguished by observing their features through a light microscope. Such features include length, position of a constricted region (centromere), and the particular patterns of light and dark bands that result from treatment with stains.

Genes are chromosome pieces whose base sequence (e.g., ATTCGGA) determines how, when, and where the body makes each of the many thousands of different proteins required for life. Humans have an estimated 25,000 genes, with an average length of about 3000 bases. Genes make up less than 2% of human DNA; the remaining DNA has important but still unknown functions that may include regulating genes and maintaining the chromosome structure.

Researchers hunt for disease-associated genes by looking for base changes found only in the DNA of affected individuals. Numerous disorders and traits mapped to particular chromosomes are displayed on this poster. Some disorders, such as cystic fibrosis (chr. 7) and sickle cell anemia (chr. 11), are caused by base sequence changes in a single gene. Many common diseases such as diabetes, hypertension, deafness, and cancers have more complex causes that may be a combination of sequence variations in several genes on different chromosomes, in addition to environmental factors.

In April 2003, scientists in the international **Human Genome Project (HGP)** completed a highly accurate, 3-billion base pair human genome reference DNA sequence. The completion of the HGP coincided with the 50th anniversary of the publication of the structure of the DNA molecule by James Watson and Francis Crick. This achievement earned them the Nobel Prize and spawned the new field of molecular biology that led to the inception of the HGP in 1990. DNA sequences are being obtained for the genomes of many other organisms as well and are critical for comparative studies leading to a greater understanding of human biology and that of all living organisms.

A New Era of Customized Medicine. Knowing the DNA sequence is important because it affects responses to particular medicines, resistance to infections and toxins, and it may even influence behavior. Sequence variations also can cause or contribute to such disorders as those named on this poster.

These new data and powerful DNA analysis tools will usher in a new era of medicine that could allow doctors to detect disease at earlier stages, make more accurate diagnoses, and customize drugs and other medical treatments to fit an individual's own DNA sequence. The eventual understanding of gene functions will lead to more focused and effective treatments with fewer side effects.

For More Information

- Human Genome Project Information: Comprehensive HGP information and a look at the "new biology" of the 21st century
  www.ornl.gov/hgmis/home.shtml
- Genomics and Its Impact on Science and Society: A Primer
  www.ornl.gov/hgmis/publicat/primer/
- Genome Education Resources
  www.ornl.gov/hgmis/education/education.shtml
- DOE Genomics:GTL: Explores how microbial and plant genomes function for bioenergy and other applications
  http://genomicsgtl.energy.gov
- Medicine and the New Genetics: How genetic technologies will revolutionize medicine
  www.ornl.gov/hgmis/medicine/medicine.shtml
- Nature Human Genome Collection: Detailed analyses of all the chromosomes
  www.nature.com/nature/supplements/collections/humangenome
- National Human Genome Research Institute: National Institutes of Health genome program
  www.nhgri.nih.gov
- Careers in Genetics and the Biosciences: Resources for students and teachers
  www.ornl.gov/hgmis/education/careers.shtml
- DOE Joint Genome Institute: Facility for integrated high-throughput sequencing and computational analysis
  http://jgi.doe.gov
- Ethical, Legal, and Social Issues: Implications surrounding use of genome data
  www.ornl.gov/hgmis/elsi/elsi.shtml

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Free print copy: 865.574.0597 or
www.ornl.gov/hgmis/posters/chromosome

Explore the Human Genome Online!

Gene Gateway

www.ornl.gov/hgmis/posters/chromosome

Step-by-step instructions for using the Web to learn about:

Genetic Disorders

- Causes, inheritance, symptoms, diagnosis, treatments
- Associated genes
- Support groups and organizations
- Genetic health professionals
- Articles and other materials

Genes and Proteins

- Gene name, symbol, size, protein product
- Chromosome maps
- Gene and protein sequence data
- Similar sequences in other organisms
- Gene mutations associated with disorders
- Molecular structures of proteins

Legend

- Regions reflecting the unique patterns of light and dark bands seen on human chomosomes stained to allow viewing through a light microscope.
- The centromere, or constricted portion, of each chromosome.
- Chromosomal regions that vary in staining intensity and sometimes are called heterochromatin (meaning "different color").
- Variable regions, called stalks, that connect a very small chromosome arm (a "satellite") to the chromosome.

Information Sources

Genes associated with the disorders and other traits listed on this poster were selected from *Online Mendelian Inheritance in Man (OMIM)*, which designated the status of each of these as confirmed or provisional as of July 2000. Listing of genes on Y chromosome updated November 2002. The number of base pairs reported for each chromosome is based on finished human genome sequence data from the National Center for Biotechnology Information Build 34, Version 2, accessed February 4, 2004 (www.ncbi.nlm.nih.gov/genome/guide/human/).

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