Exome

The exome is the part of the genome formed by exons, the sequences which when transcribed remain within the mature RNA after introns are removed by RNA splicing. It consists of all DNA that is transcribed into mature RNA in cells of any type as distinct from the transcriptome, which is the RNA that has been transcribed only in a specific cell population. The exome of the human genome consists of roughly 180,000 exons constituting about 1% of the total genome, or about 30 megabases of DNA.

Though comprising a very small fraction of the genome, mutations in the exome are thought to harbor 85% of mutations that have a large effect on disease.

Exome sequencing has proved to be an efficient strategy to determine the genetic basis of more than two dozen Mendelian or single gene disorders.

Examples of research projects using exome sequencing include the nonprofit Personal Genome Project (PGP), the nonprofit Rare Genomics Institute (RGI), the NIH-funded Exome Project, the NHGRI-funded Mendelian Exome Project, the NHLBI Grand Opportunity Exome Sequencing Project and the microarray-based Nimblegen SeqCap EZ Exome from Roche Applied Science.