Annotation and Functional Analysis of Human Chromosome 7 for Disease Research

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Our group has worked to describe and understand human chromosome 7 as a discrete organelle affecting both the inheritance and the expression of genes in health and disease. We are incorporating all biological information (called process called annotation) around the DNA sequence and are working to complete a fully integrated genetic, physical, and gene map for this portion (5%) of the human genome. We are also characterizing all clinical and functional information relevant to chromosome 7. This strategy has already allowed us to so far identify around 2000 genes and transcription units. Through collaborations with others we have also identified the genes that cause holoprosencephaly (SHH), distal tubular renal acidosis (ATP61B), type II citrullinemia (CTLN2), sacral agenesis and Currarino syndrome (HLXB9), splenic lymphoma (CDK6), hereditary papillary renal carcinoma (MET) and other diseases. Moreover, the phenomena of position effect and genomic polymorphism mutations could be established as contributing mechanisms in human disease. We are now comparing the DNA sequence of chromosome 7 to that of other human chromosomes, as well as to the genomes of other species. This comparative annotation is providing clues into the origins and evolution of chromosome 7 and its role in human development.