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and how new molecular and cellular approaches including cell atlases and organoids are enabling exploration of the candidate causal factors that underlie human specific traits special issue on genetic epidemiology of complex diseases impact of population history and modelling assumptions volume 138 january december 2019 jan dec 2019 issue 11 12 december 2019 human genetics focuses on publishing timely articles covering all aspects of human genetics it covers a broad range of topics from gene structure and papers may be considered for publication in the sections listed below original investigations original investigations are definitive and comprehensive descriptions of major research findings which are of broad significance for readers of human genetics papers may be presented as full articles or short communications we have particular interest in the areas of genomic technology genome structure and function genetic modification human variation and population genetics human evolution and importantly all aspects of human genetic disease and including individualized medicine recent remarkable advances in high throughput genotyping and next generation dna sequencing technologies have generated massive human genome sequencing data in this review we provide an overview of the types of molecular change that have occurred during human evolution as revealed by comparative genomics across the great apes and studies of ancient dna from archaic hominins highlighting molecular changes linked to human specific traits in this special issue we compile an

exciting group of comprehensive reviews from diverse perspectives on topics ranging from functional non coding variant prediction and annotation to discovery and validation of disease associated loci and genes to document the innovation recent progress and potential biomedical impact of computational and human molecular genetics and genomics the discovery of genes responsible for more than 5000 rare mendelian diseases has facilitated genetic diagnostics for many patients pregnancy related molecular genetics genomic medicine is a broad scope journal for medical genetics basic human genetics research including genomic variation inherited disorders abstract background the triggering receptor expressed on myeloid cells 2 protein trem2 plays a crucial role in various biological processes including osteoclast differentiation supporting human genetic evidence for new drugs approved by the fda in 2021 availability of genetic evidence implicating each of the 50 drug targets columns as likely causal genes for the this chapter provides a review of the basic principles of human genetics including molecular genetics cytogenetics genetic transmission and genomics it then provides an introduction to the principles of genetic testing and major approaches to the treatment of genetic disorders the annual review of genomics and human genetics is a peer reviewed scientific journal published by annual reviews since 2000 it releases an annual volume of review articles relevant to the fields of genomics and human genetics aravinda chakravarti and eric d green have been the journal s

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