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ISSN: 2161-1041
Hereditary Genetics: Current Research

Opinion Article

Emerging Perspectives on Hereditary Diseases: Advances in Genetic Research and Precision Medicine

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DESCRIPTION

Hereditary diseases are disorders that are transmitted from one generation to another through genetic material. These conditions arise from pathogenic variants in genes or chromosomes that are inherited from one or both parents. Hereditary disorders encompass a broad spectrum of conditions, including single-gene disorders, multifactorial diseases, mitochondrial disorders and chromosomal syndromes. Recent developments in molecular genetics have transformed the understanding of hereditary diseases by enabling earlier diagnosis, improved disease classification and personalized therapeutic strategies.

Single-gene hereditary diseases result from mutations in a single gene and follow Mendelian inheritance patterns, including autosomal dominant, autosomal recessive and X-linked inheritance. Disorders such as cystic fibrosis, caused by pathogenic variants in the *CFTR* gene, Huntington disease associated with expansions in the *HTT* gene and sickle cell disease resulting from mutations in the *HBB* gene illustrate how alterations in individual genes can lead to severe clinical manifestations.

Multifactorial hereditary diseases represent another important category in which multiple genes interact with environmental and lifestyle factors. Conditions including cardiovascular disease, type 2 diabetes mellitus, hypertension and certain cancers exhibit hereditary susceptibility while also being influenced by diet, physical activity, environmental exposures and aging. Genome-Wide Association Studies (GWAS) have identified numerous susceptibility loci that contribute to disease risk, allowing researchers to better understand the biological pathways involved in disease development.

Current research has emphasized the role of precision medicine in hereditary disease management. Precision medicine integrates genomic information with clinical and environmental data to tailor prevention, diagnosis and treatment strategies to individual patients. Whole Genome Sequencing (WGS) and Whole Exome Sequencing (WES) have become valuable diagnostic tools, particularly for patients with rare hereditary disorders whose conditions remain undiagnosed through conventional clinical evaluation.

Gene therapy has emerged as one of the most promising therapeutic approaches for hereditary diseases. Instead of treating symptoms alone, gene therapy aims to correct or replace defective genes responsible for disease. Clustered Regularly Interspaced Short Palindromic Repeats (CRISPR)-associated protein 9 (CRISPR-Cas9) genome editing technology has demonstrated remarkable potential in correcting pathogenic mutations at their genomic source. Experimental studies targeting inherited blood disorders, retinal diseases and neuromuscular conditions have produced encouraging clinical outcomes. Although long-term safety and ethical considerations remain under investigation, gene-editing technologies continue to advance rapidly.

Prenatal and preimplantation genetic testing have expanded reproductive options for families with hereditary disorders. Non-Invasive Prenatal Testing (NIPT), chorionic villus sampling, amniocentesis and Preimplantation Genetic Testing for Monogenic Disorders (PGT-M) enable early detection of inherited genetic abnormalities before or during pregnancy. These technologies provide prospective parents with valuable information for reproductive decision-making while supporting early clinical intervention when appropriate.

Epigenetics has introduced additional complexity to hereditary disease research. Epigenetic mechanisms, including DNA methylation, histone modification and non-coding ribonucleic Acid (RNA) regulation, influence gene expression without altering the underlying DNA sequence. Current studies suggest that epigenetic modifications contribute to disease onset, progression and variable clinical presentation among individuals

In conclusion, hereditary disease research has entered an era of unprecedented scientific progress driven by advances in sequencing technologies, precision medicine, artificial

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Received: 23-May-2026, Manuscript No. HGCR-26-32037; **Editor assigned:** 25-May-2026, PreQC No. HGCR-26-32037 (PQ); **Reviewed:** 09-Jun-2026, QC No. HGCR-26-32037; **Revised:** 16- Jun -2026, Manuscript No. HGCR-26-32037 (R); **Published:** 23-Jun-2026, DOI: 10.35248/2161-1041.26.15.308

Citation: Moreau O. (2026). Emerging Perspectives on Hereditary Diseases: Advances in Genetic Research and Precision Medicine. Hereditary Genet. 15:308.

intelligence, pharmacogenomics, gene therapy and population genomics. These innovations are expected to transform hereditary disease management while improving patient outcomes and supporting the development of truly personalized healthcare.