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Commentary

What is Monogenic Disorders?

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DESCRIPTION

The human genome, which contains the entire deoxyribonucleic acid (DNA), can be thought of as the model for all cellular structures and activities. Our genome is believed to contain 25,000 protein-coding genes, and knowing their sequences can help match different disease phenotypes to their respective genes.

Human genetic diseases are classified into five types: single gene disorders (characterized by mutations at individual loci), multifactorial and polygenic disorders (characterized by the interaction of multiple genes, sometimes in concert with environmental factors), chromosomal abnormalities, mitochondrial inheritance, and chromosomal abnormalities.

When a single gene is identified as the cause of a disease, it is called a single-gene disorder or a Mend Elian disorder. The disease caused by a single genetic mutation can manifest itself in several ways. They are classified according to whether the trait is sex-specific (usually X-linked) or not (auto chromosomal). Monogenic disorders (also called Mend Elian diseases or traits) are generally rare.

The onset of disease due to a single genetic mutation can occur in several major patterns or modes. They are grouped according to whether their characteristics are gender specific (usually Xlinked) or not (autosomal). As a rule, monogenic disorders (also called traits or Mendelian diseases) are relatively rare.

Monsoonal dominant disorders usually occur in individuals who contain a single mutated copy of a disease-related gene. Affected individuals are heterozygous for the gene, which means that inheriting a single copy from the affected mother or affected father is sufficient to cause the disease; therefore, the presence of a non-mutant.

Another common mode of transmission is a single autosomal recessive disorder, in which two copies of the mutated gene are needed to develop the disorder. They inherit one allele from the mother and one from the father, the risk of transmission is 25%, while half of the unaffected children will carry the X-linked gene inherited in a similar fashion to dominant inheritance.

The chromosome usually excludes more women affected than men, although these disorders are rare. X-linked recessive conditions usually occur only in males, as the female second chromosome provides the normal allele, but males who inherit the recessive gene on their own X chromosome are affected. Extremely rare Y-linked monogenic diseases are still passed from affected fathers to their sons.

CONCLUSION

Most of the monogenic disorders are uncommon to show up even their reasons are nevertheless unknown while some are pretty not an unusual place with recognised reasons. Various diagnostic techniques and remedy techniques were advanced for them which have now no longer been proved sufficient to deal with the issues by conventional approaches. Therefore, a few latest and updated approaches also are being carried out inside the area of remedy of these issues that consists of Gene Therapy, Stem cell transplantation and Bone Marrow Transplant.

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