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Aicardi Syndrome is Frequently Fatal in Males

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Editorial

A chromosomal anomaly or a mutation in a single gene (monogenic) or numerous genes (polygenic) might cause it. Although polygenic illnesses are the most frequent, the term is typically applied to conditions that have a single genetic origin, such as a mutation in a gene or chromosome. A genetic disorder is a medical condition caused by one or more genetic defects. Some illnesses have X-linked inheritance and are caused by a mutation on the X chromosome. Only a few diseases are passed on through the Y chromosome or mitochondrial DNA (due to their size). There are about 6,000 documented genetic illnesses, and new genetic abnormalities are described in the medical literature on a regular basis. There are around 600 genetic illnesses that can be treated. A known single-gene problem affects about 1 in 50 persons, while a chromosomal disorder affects about 1 in 263 people. As a result of congenital genetic mutations, around 65 % of people experience some sort of health issue [1, 2].

Because of the high number of genetic abnormalities, one out of every twenty-one people is affected with a "rare" genetic disorder (usually defined as affecting less than 1 in 2,000 people). The majority of genetic illnesses are uncommon in and of themselves. Birth defects can be caused by genetic abnormalities that exist before birth, and some genetic disorders cause birth deformities, but birth defects can also be caused by developmental rather than hereditary factors. An acquired disease is the polar opposite of a hereditary sickness. Most cancers are acquired diseases, despite the fact that they contain genetic abnormalities in a small percentage of the body's cells. However, some cancer syndromes are hereditary genetic illnesses, such as BRCA mutations. The outcome of a single mutated gene is a single-gene disorder (or monogenic disorder). Single-gene diseases can be passed down the generations in a variety of ways. However, genetic imprinting and uniparental disomy may have an impact on inheritance patterns. Although the distinctions between autosomal and X-linked types are clear, the distinctions between recessive and dominant types are not (since the latter types are distinguished purely based on the chromosomal location of the gene). For example, achondroplasia, the most prevalent form of dwarfism, is commonly thought to be a dominant illness, however children with two achondroplasia genes have a severe and usually fatal bone disorder, which achondroplasics could be carriers for.

Although sickle cell anaemia is a recessive condition, heterozygous carriers have greater malaria resistance in early childhood, which might be considered a related dominant condition. When a couple with one or both partners suffering from or carriers of a single-gene disorder wants to start a family, they can use in vitro fertilisation, which allows for preimplantation genetic testing to see if the embryo has the genetic disorder. Single-gene abnormalities cause the majority of congenital metabolic diseases, often known as inborn errors of metabolism. Because many single-gene flaws can reduce a person's fitness, they are found in the population at lower frequencies than would be expected based on simple probability calculations. To be afflicted by an autosomal dominant condition, a person only needs one mutant copy of the gene. Each affected person usually has one parent who is also affected. Normally, neither parent with a faulty gene exhibits symptoms. With each pregnancy, two unaffected people who each possess one copy of the mutant gene have a 25% chance of conceiving a kid with the condition. Albinism, medium-chain acyl-CoA dehydrogenase deficiency, cystic fibrosis, sickle cell disease, Tay–Sachs disease, Niemann–Pick disease, spinal muscular atrophy, and Roberts syndrome are examples of this type of condition. Other traits, such as wet vs. dry earwax, are determined in an autosomal recessive manner as well. Some autosomal recessive disorders are frequent because harbouring one of the defective genes formerly provided a little amount of protection against infectious diseases or toxins like tuberculosis or malaria. Mutations in genes on the X chromosome cause X-linked dominant diseases.

This inheritance pattern is found in just a few illnesses, one of which being X-linked hypophosphatemic rickets. Both males and females are affected by these illnesses, with males suffering from them more severely than females. Because some X-linked dominant disorders, such as Rett syndrome, incontinent pigment type 2, and Aicardi syndrome, are frequently fatal in males in gestation or shortly after birth, they are mostly found in girls. Extremely uncommon cases of boys with Klinefelter syndrome (44+xxy) inheriting an X-linked dominant illness and exhibiting symptoms more similar to those of a female in terms of disease severity are exceptions to this result. Men and women have different chances of passing on an X-linked dominant disease. Because they inherit their father's Y chromosome, the boys of a man with an X-linked dominant illness will be unaffected, but his daughters will all acquire the disorder [3-5].

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Conflict of Interest

The Author declares there is no conflict of interest associated with this manuscript.

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