Special Features

Common genetic disorders in India

Dr V L Ram Prasad
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India represents one-sixth of the world population and consists of ethnically, geographically and genetically diverse populations with several thousand endogamous groups. The load of genetic disorder is relatively high in some Indian communities due to consanguineous marriage practiced in those communities. Most common genetic disorders due to such wedlock are 6PD deficiencies, Down syndrome, Sickle cell disease and ß-thalassemia etc. These genetic disorders happen when a particular gene is dominant or when a mutation is present in both copies of a recessive gene pair. A gene itself is not functional, but expressed as a protein which is the functional unit of that gene. Thus, set of genes, called as genome, contains all the requisite genetic instructions for developing and directing the activities within an organism and are inherited from parents. They encode the information required for every biological function. Each gene is made of four chemical bases in a sequential manner. The genomic sequence, often referred as DNA of every individual is unique as the structure/sequence of gene/genome slightly varies between individuals. These small/large variations, i.e. mutations, in a gene or across genome may affect the function of its encoded protein products and sometimes may cause a disease. Different mutations cause different disorders giving rise to what is medically termed as clinical or genetic disorders.

Some of the common genetic disorders in India are: Down Syndrome; Thalassemia; Sickle Cell Anemia; Cystic Fibrosis, and Tay-Sachs.

Down Syndrome, also, known as Trisomy 21, is a chromosomal disorder caused because of an abnormal cell division resulting in an additional copy of chromosome 21. Some of the key characteristics of Down syndrome include distinct facial appearance, intellectual disability, developmental delays and may be associated with thyroid or heart disease. It is a rare genetic disorder occurring in about 23000 to 29000 children per year in India. Additional features of this syndrome may include slurry speech and hearing & visual flaws.

Thalassemia is a genetic disorder related to blood which involves the body's inability to make the required amount of haemoglobin (found in red blood cells) which helps carry oxygen to the entire body. This makes the person extremely anaemic resulting in pale skin, fatigue, and a host of other serious complications like increased risk of developing abnormal blood clots. There are two types of Thalassemia viz; Alpha thalassemia and ß-thalassemia, the latter being of a more serious nature and also referred to as Cooley's anaemia. A person with ß-thalassemia shows symptoms as early as two years after birth in the form of paleness of the skin, irritability, poor appetite, and very slow growth. Proper treatment includes routine blood transfusions and other therapies.
Alpha thalassemia: There are two types of alpha thalassemia. Major one is an extremely serious form of the disease. Most individuals with this condition die before or shortly after birth. Pregnant women who carry foetuses infected with this type of thalassemia are subject to very serious complications which may affect them during the pregnancy and delivery phases. Another type of alpha thalassemia is the haemoglobin H disease which in turn occurs in different ways.

All various types of Thalassemia have serious repercussions which are best described by a reduced production of normal haemoglobin or its complete absence.

Sickle cell anaemia is an inherited disorder which affects the red disc-shaped red blood cells. Once affected these cells take on an abnormal shape – that of a sickle/crescent which split with ease resulting in many consequences including early death of red blood cells leaving a shortage of healthy red blood cells (sickle cell anaemia) and blockage of blood flow causing pain (sickle cell crisis) anaemic conditions. The latter is due to their shape which hinders their movement through small blood vessels. As a result, they get stuck in the small blood vessels and prevent the flow of blood from reaching the different parts of the body and thus cause pain.

The life span of these cells varies between 10-20 days which is a huge departure from the normal lifespan of 120 days. This state of a limited life span causes the person prone to infections and results in permanent harm to vital organs like the heart, cerebellum, lungs, kidneys, as well as other parts of the body.

The first signs of sickle cell anaemia are depicted through experiencing a sharp pain in the hands and feet. Other symptoms of Sickle cell anaemia include high levels of fatigue, irritation, jaundice, regular infections and even bed-wetting. Babies affected by SCA tend to display high levels of irritability as well. Sickle cell anaemia comes in varying types but the symptoms remain the same albeit with varying degrees of severity.

Cystic fibrosis is an inherited life-threatening genetic disorder of the mucus and sweat glands. This is caused due to mutations in a single gene on Chromosome 7. The gene, Cystic fibrosis transmembrane conductance regulator, encodes a protein that functions as a membrane channel to transport chloride ions into and out of cells that produce mucus, sweat, saliva, tears, and digestive enzymes. But, the mutations in that gene hinders the transmembrane functioning of chloride channel. Thus, cystic fibrosis affects the sodium channels in the body, causes your mucus to be thick and sticky. The mucus clogs the lungs, causing breathing problems and making it easy for bacteria to grow. This can lead to repeated lung infections and lung damage. Over a period, weakens a person's ability to breathe. Cystic fibrosis occurs in both, males and females and research has proved that 1 in every 25 people suffer from it but depict no symptoms. This disease is often diagnosed at birth but is not contagious.

Tay-Sachs is a rare genetic disorder which affects the nervous system. The characteristics of this disorder is progressive deterioration of nerve cells and of mental and physical abilities that begins around six months of age. Though its progress cannot be hindered, genetic testing conducted in a child while it's still in a mother's womb & helps to understand whether the child is affected or not. Tay-Sachs can be diagnosed in the early stages of pregnancy either through amniocentesis or through blood tests.

Tay-Sachs disease results from defects in a gene on chromosome 15 that codes for production of the enzyme hexosaminidase A (Hex-A). This defective gene prevents the body from making requisite amounts of this enzyme causing gangliosides (chemicals) to build up in the brain cells, leading to their complete destruction, affecting the child's mental and physical development. If a child gets 2 copies of this defective gene each from one parent, he gets affected. Tragically, this disease damages the central nervous system of a child while still in the mother's womb and the symptoms are visible between 12 to 24th week of pregnancy. An incurable disease, its symptoms include blindness, deafness, seizures, and decrease in muscle tone, slow growth and loss of a muscle function leading to paralysis.

However, if the child gets only one copy of the defective gene from either of the parents, he will not be affected because the other copy is healthy. But, that child will be a carrier and may pass on the faulty gene to their children.

Management of genetic disorders
Control and management of the genetic disorders depend on identification of the variants in the genome that are causally linked with the disease. The need of the hour to improvise the situation is integration of genetic services into primary health medical services and educating communities of the aftermath of consanguineous marriages. Appropriate genetic counselling by trained physicians, increasing the number of departments of medical genetics in medical schools will help increase an awareness of the various repercussions and give an insight to communities largely affected by this.

Research has proved that at least 70% of birth defects can be prevented if the evidence based community genetics services are made available. Tragically, religious and cultural aspects of communities in India are primarily responsible for many birth defects which occur in our country. Prenatal genetic testing can help detect symptoms of genetic disorders. This assists the concerned parents to choose the correct option for a probable course of medication to arrest, prevent its progress or management of its consequences. In the absence of a suitable cure, these tests can guide them to make a decision on the next best alternative to avail of. For example, early detection of Down syndrome, combined with guidance and education as well as various services can help improvise the situation of such kids and help them deal with it bravely. With the correct preventive measures, genetic testing and screening, the existing state of birth defects can be controlled and managed effectively even if at a slow pace.

(Author is COO of MedGenome)

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