Health Files
Dialogues by Health Gurus

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Deaf blindness Awareness Week – Can genetics help?

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"Blindness separates people from things, deafness separates people from people." - Helen Keller

The last week of June was celebrated as Deaf blindness Awareness Week, as is every year, to commemorate the birth anniversary on June 27th, of Helen Keller – a deaf-blind American woman, a pioneer for deaf blindness awareness, who symbolized the achievements of deaf blind people across the world.

Deaf blindness is a syndrome characterised by a combined sight and hearing impairment that causes difficulties with communication, access to information and mobility. Otherwise known as dual sensory loss, it is often researched as distinct sub groups: those with congenital vision and hearing impairment and those with acquired dual sensory impairment - because of their different developmental conditions. Patients suffering from this generally fall into 4 groups – congenitally deafblind, congenitally deaf and acquired blindness later in life, congenitally blind and acquired deafness later in life, and acquired deafblindness later in life.

The incidence of deaf blindness is very low, and the data on exact numbers are not very well known. The generally accepted estimates are that approximately 10% of the general population has a hearing loss, out of which approximately 1% is also blind or has a serious loss of vision. There are an estimated 40,000-70,000 deafblind people living in the US.

There is no data available regarding the size of the deaf blind population in India. As of date, there has been no comprehensive study or research done to determine the true incidence of deafblindness. Estimates, based on information gathered from community-based projects, indicate that there could be more than 400,000 deafblind people in our country. Overall we can predict that 0.04% of general population have deafblindness as a disability.

GENETICS OF DEAF BLINDNESS

The primary causes of vision and hearing loss are – prenatal viral/bacterial/chemical impacts, postnatal injuries, and hereditary/chromosomal disorders. One of the main causes of deafblindness is Rubella, causing Congenital Rubella Syndrome, or cytomegalovirus infection -bothcontracted by the pregnant mother. There are also hereditary syndromes associated with deafblindness, of which Usher's syndrome is of prime importance, and is thought to be responsible for 3 percent to 6 percent of all childhood deafness and about 50 % of deaf blindness in adults.

Characterised by retinitis pigmentosa and hearing loss, Usher's syndrome is an autosomal recessive disorder. Researchers have identified three major types of Usher syndrome, designated as types I, II, and III. These types are distinguished by their severity and the age at which signs and symptoms appear.

In addition to this, non-syndromic deafness can also be inherited in autosomal dominant (designated DFNA) or recessive (designated DFNB) patterns, and is very complex in nature. Over 100 loci and 46 nuclear genes have been associated with it.

The genomic services industry has facilitated the development of diagnostic tests for such hereditary syndromes, whereby they can be identified and appropriate therapy defined. Published literature on genes implicated in Usher's syndrome, a key condition leading to deafblind condition, allows development of disease specific genetic testing panels. Although interpretation of deafblind behaviour and care-giving for individuals affected by it poses a significant challenge, timely and accurate diagnosis can be very useful in helping decide a treatment protocol to enable the individual's maximal participation in society.

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