Health Files
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Next Generation Sequencing for the diagnosis of Congenital Cataract

14 Aug 2015

August is National Cataract Awareness Month. Cataract, which can be defined as any opacity of the crystalline lens, results when the refractive index of the lens varies significantly over distances approximating the wavelength of the transmitted light. This variation in the refractive index can result from changes in lens cell structure, lens protein constituents, or both. Cataract development may be influenced by factors like over production of oxidants, or smoking. They are often categorised by the reason for formation - age-related cataracts , congenital cataracts (these are often hereditary) , secondary cataracts (possibly caused by disease/medications) , traumatic cataracts (from injury to the eye), or from radiation (after radiation therapy for cancer).

GENETICS OF CATARACTS

Age-related cataracts develop when progressive opacities begin to form in the lens around the onset of middle age. Lens proteins are known to undergo a wide variety

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Congenital cataract is one of the major causes of childhood blindness in India. Approximately 25% of non-syndromic cataracts are inherited. Hereditary cataracts are estimated to account for between 8.3% and 25% of congenital cataracts. The lens alone may be involved, or lens opacities may be associated with other ocular afflictions, such as microphthalmia, aniridia or retinal degenerations. Cataracts may also be part of multisystem genetic disorders, such as chromosome abnormalities, Lowe syndrome, neurofibromatosis type 2, Down's syndrome, Wilson's disease or myotonic dystrophy. In some cases this distinction is blurred. Inherited cataracts may be isolated in some individuals and associated with additional findings in others, as in the developmental abnormality anterior segment mesenchymal dysgenesis, resulting from abnormalities in the PITX3 gene.

Congenital cataracts are also genetically heterogeneous. It is known that different mutations in the same gene can cause similar cataract patterns, while the highly variable morphologies of cataracts within some families suggest that the same mutation in a single gene can lead to different phenotypes. To date, more than 25 loci and genes on different chromosomes have been associated with congenital cataract. Mutations in distinct genes, which encode the main cytoplasmic proteins of human lens, have been associated with cataracts of various morphologies, including genes encoding crystallins, lens specific connexins, major intrinsic protein or aquaporine, cytoskeletal structural proteins, paired-like homeodomain transcription factor 3, avian musculoaponeurotic fibrosarcoma and heat shock transcription factor 4.

GENOMICS DIAGNOSTICS

Numerous genes have been implicated in the development of congenital cataracts. Some of the genes are EPHA2 , GJA8 ,CRYGD ,CRYGC ,CRYGB ,CRYBA2 ,FYCO1 ,BFSP2 ,CRYGS and WFS1, among others. Genomics diagnostics companies offer panels that screen for mutations in the genes and provide necessary insights to the doctors. It is a very useful tool in the clinical practice as congenital cataract can lead to permanent blindness, if unaddressed. Prompt diagnosis and timely treatment can prevent this and hence improve the life of the patients.

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Hiranjith is associated with MedGenome, the first mover and market leader in genomic-based diagnostics and research from India.

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