Atypical morphological variants of congenital cataract in Down's syndrome

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DESCRIPTION

A myriad of ocular disorders are associated with Down's syndrome including refractive errors, strabismus and cataract resulting in significant visual impairment. Haargaard *et al* estimated a population-based frequency of 1.4% of early cataract in Down's syndrome with less than 1% requiring surgical intervention.¹ Morphological variants described in Down's syndrome are punctate cortical opacities sparing the nucleus, zonular, cortical cataract, with total cataract being the most common.^{1 2} We describe few rare congenital cataract morphology observed in two patients with Down's syndrome.

A 2-month-old male child presented with unilateral partially absorbed membranous cataract with dense white anterior capsular plaque (ACP) (*figure 1A*). The thick plaque was well appreciated on ultrasound biomicroscopy (UBM) at a frequency of 35 MHz as a thick hyperechoic linear line on anterior capsule with loss of lens volume (*figure 1B*). The histopathology examination of the ACP revealed increased subepithelial fibrosis (*figure 1C*).

A 2-year-old male child presented with total intumescent cataract with dense ACP Oculus dexter (OD) and a fan-shaped cataract involving the visual axis oculus sinister (OS) (*figure 2A,C*) better appreciated on UBM (*figure 2B,D*).

Figure 1 Clinical photograph of membranous cataract in a 2-month-old child with Down's syndrome with thick anterior capsular plaque at the centre (red arrow, A), detected by a dense homogeneous hyperechoic linear line on ultrasound biomicroscopy (red arrow, B). Histopathological examination under H&E stain of the capsular plaque depicted the anterior cuboidal epithelium cells of the anterior capsule (black arrow, C) with increase in extracellular matrix and fibroblast deposition (white arrow, C).

Plaques are formed when lens epithelial cells (LECs) undergo epithelial mesenchymal transdifferentiation (EMT) into myofibroblasts and secrete extracellular matrix.^{3 4}

It takes place in two phases—proliferative phase where cells undergo rapid proliferation and trans-differentiation phase secreting extracellular matrix. Occurrence of ACP in paediatric patients may be due to the high proliferation capacity of paediatric LECs.⁵ The occurrence of ACP has been found to be more in eyes with mature cataract.⁵

Fan-shaped cataract is a rare finding, reported in a previous paper with an autosomal dominant inheritance in association with microcornea. This characteristic phenotype was linked to the mutation in α -crystallin (CRYAA) gene.⁶ The α -crystallins comprise up to 40% of lens protein, acting as molecular chaperones facilitating proper folding of other lens protein and maintaining proper functional state.⁷ A fan-shaped nuclear opacification has been reported in another paper with an X-linked inheritance with genetic locus on chromosome Xp22.⁸

There are about 45 genetic loci at present to which isolated cataracts have been mapped with 38 specific genes known in inherited cataracts.⁹ The gene found in this Indian family with fan-shaped cataract has a

Figure 2 Clinical photograph of total intumescent cataract with thick dense anterior capsular plaque (white arrow, A) with homogeneous hyperechoic linear line corroborating to the plaque on ultrasound biomicroscopy (UBM) (white arrow, B). Clinical photograph of a typical fan-shaped cataract with three radial spokes of triangular-shaped nuclear opacification radiating from a central zonular opacification in a 2-year-old child with Down's syndrome (C). UBM depicts the central opacification of the lens (D).

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genetic locus on chromosome 21 and Down's syndrome is caused by trisomy 21, a coincidental observation.

To the best of our knowledge, ACPs and fan-shaped congenital cataracts have not been reported in association with Down's syndrome.

Patient's perspective

If more awareness was created at the time of birth of my child, I would have brought for routine ophthalmological screening earlier.

Learning points

- Periodic ophthalmic evaluation is recommended since early childhood in Down's syndrome for early detection of congenital cataract and other ophthalmic disorders.
- Ultrasound biomicroscopy imaging enables better appreciation of the morphological anomalies prior to surgery, hence enabling better surgical planning.
- Anterior capsular plaque (ACP) in paediatric population can occur even without trauma or any other component of ocular inflammation.
- Presence of dense ACP in Down's syndrome depicts the long-standing proliferation and transdifferentiation changes occurring in lens epithelial cells.

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Disclaimer Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research. They should not be used in isolation to guide treatment choices or public health policy.

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