IMI Management and Investigation of High Myopia in Infants and Young Children

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Prevalence of high myopia in infants and young children
High myopia in children under 5-6 years of age is rare. In China and Singapore, prevalence rates of 0.03-0.2% have been reported for myopia worse than -6.0D in children under 7 years of age. In the USA, the prevalence of myopia worse than -4.0D has been reported to range from 0.6-0.8% in 5–6-year-olds.

Etiology
The environmental and genetic risk factors for high myopia in this younger population are distinct from those reported for school aged children. One of the more notable environmental factors is premature birth, particularly in children who develop Retinopathy of Prematurity (ROP). There are two main types of genetic etiologies. The first one involves the interaction between numerous known genetic risk factors and environmental factors like near work and outdoor exposure. The second occurs due to mutations in a single gene that significantly affects refractive development. This type of myopia is known as monogenic high myopia and development is largely independent of common environmental risk factors. Monogenic high myopia can occur on its own or be associated with various eye and non-eye related features, which is referred to as syndromic myopia.

Monogenic forms of myopia can be broadly categorized into four groups:
(1) ametropic retinal dystrophies, e.g., genes associated with cone-rod dystrophies, albinism, retinitis pigmentosa
(2) connective tissue disorders, e.g., Stickler Syndrome, Marfan syndrome, Ehlers-Danlos syndrome
(3) monogenic isolated high myopia
(4) other disorders e.g., causing corneal or lens malformations, congenital glaucoma

Clinical evaluation
History taking needs to be tailored to identify possible monogenic inheritance patterns and the symptoms of the most significant syndromic forms of myopia. Birth history is important in relation to the possible contribution of prematurity, and assessment of milestones provides a simple test for developmental delay which is a common finding in childhood high myopia.

General clinical evaluation of general psychomotor development, facial morphology, and limbs can indicate the need for review by a pediatrician or clinical geneticist.

Visual acuity and colour vision evaluation can differentiate children with retinal dystrophy.

Pupil reactions, slit lamp and retinal examination can identify features of retinal disease, ROP or connective tissue disorders.

Intraocular pressure in an infant is needed to exclude the possibility of congenital or early onset glaucoma.

Ocular biometry is essential as there are many conditions such as ROP and Keratoconus where the associated myopia is not primarily axial.

Ocular imaging such as wide-angle fundus photography, fundus autofluorescence imaging, optical coherence tomography (OCT), and optical coherence tomography angiography, can all provide critical diagnostic information regarding possible inherited retinal diseases.

Electrophysiology is used to help diagnose a variety of visual disorders and are essential for detecting retinal dystrophies.

Role of primary eye care practitioners
Once the initial diagnosis of high myopia is made and optical correction provided, the priority is to determine whether there is an associated systemic or ocular disorder. The primary eye care community needs to be able to recognize the risk factors for syndromic forms of myopia in children so that timely and appropriate referrals for further investigation can be made when applicable. If the clinical evaluation suggests a monogenic or syndromic form of myopia, the involvement of other medical professionals may be warranted. These may include ophthalmologists who specialize in inherited disease, clinical geneticists, genetic counsellors, and/or pediatricians.

Challenges of optical correction
Optical correction needs to be optimised to avoid amblyopia and facilitate normal visual development. Although spectacles will be the primary form of optical correction, contact lenses may be more appropriate for children with significant anisometropia (such as in high anisomyopia) or where craniofacial abnormalities make the wearing of conventional spectacles challenging. Refractive surgery is also a possibility in some circumstances e.g., amblyopia unresponsive to standard therapy, non-compliance or intolerance to other optical solutions or craniofacial/orbital abnormalities making spectacles and contact lenses impractical.

Management of myopia progression
Evidence-based recommendations are difficult to provide since many of the forms of myopia described in this paper have been excluded from myopia progression trials.

Due to the different etiological factors, evidence of axial elongation should be regarded as a pre-requisite for myopia control therapy. For example, ROP and several forms of syndromic myopia tends to be corneal or lenticular rather than axial myopia.

The pattern of myopia onset and progression is often very different in syndromic myopia compared to typical myopia. In many forms of syndromic myopia, high levels of myopia are present by the age of 5 years and there is little progression thereafter.

Ensuring that there is myopia progression and axial elongation prior to intervention is warranted, especially since some myopia control treatments can have adverse effects in these cases, e.g., high dose Atropine treatment could potentially have an adverse effect on cardiac treatment for Marfan syndrome or photophobia can be exacerbated for patients with cone dystrophies. Evidence from studies on infant primates suggest caution over the use of high concentrations of Atropine for myopia control in the first year or two of life due to the risk of arrested development of the anterior segment.

When considering myopia control interventions in this group, a case-by-case approach is required. As a low-risk intervention, advice on increased outdoor activities for all children with myopia or at risk of myopia from an identified syndrome is appropriate.

Summary
- After diagnosing and correcting high myopia, it is important to determine if there is an associated medical condition that may have greater overall impact on the child's health and refer for specialized investigations and multidisciplinary evaluations where indicated.
- Biometric evaluation is essential in distinguishing between axial myopia and refractive myopia caused by abnormal development of the anterior segment of the eye and this will help determine whether myopia control intervention may be of benefit.
- Management requires careful case-by-case approach due to clinical heterogeneity and limited evidence base.

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