UNUSUAL PATTERN OF REFRACTIVE ERROR IN SIBLINGS OF LAURENCE-MOON-BIEDL SYNDROME

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ABSTRACT: Common ocular findings in Laurence-Moon-Biedl syndrome (LMBS) are rod-cone dystrophy (Atypical retinitis pigmentosa), myopia, astigmatism, anisometropia, strabismus, keratoconus, optic atrophy and nystagmus. We report three siblings presenting with decreased visual acuity due to isolated high compound myopic astigmatism.

KEYWORDS: Refractive Error.

INTRODUCTION: Laurence-Moon-Biedl Syndrome is a rare autosomal recessive disease with a spectrum of ocular associations such as rod-cone dystrophy (Atypical retinitis pigmentosa), myopia, astigmatism, anisometropia, strabismus, keratoconus, optic atrophy and nystagmus. Though myopia and astigmatism have been previously reported, this clinical image depicts the first report of such isolated high compound myopic astigmatism causing impairment in visual acuity in three siblings with Laurence-Moon-Biedl Syndrome. It also describes the available review of literature though limited due to its rarity, on the pattern of such refractive errors that may be encountered in Laurence-Moon-Biedl Syndrome.

CASE REPORT: A 7 years old Obese (BMI 22.2Kg/m2) Indian Muslim girl (Case 1) born to consanguineous parents was brought to the Ophthalmology department of a tertiary care hospital with chief complaints of gradual decrease in distant vision of three year duration. There was no history of night blindness. The parents gave a history of weight gain and intellectual impairment of 3 years duration. Systemic evaluation revealed post-axial polydactyly in both upper and lower limbs (Figure 1). Standardized formal intelligence quotient (IQ) testing revealed a mild intellectual disability. Family history was suggestive of similar complaints in her two siblings. Thyroid profile revealed borderline increase in serum TSH (6.93 uIU/ml). Ultrasonography of abdomen and pelvis revealed non-visualization of ovaries and no liver or kidney anomalies. Comprehensive ophthalmological evaluation revealed a snellen’s best corrected visual acuity (BCVA) of 6/60 in both eyes. Cover test showed alternate divergent squint with right eye dominance. Cycloplegic auto refraction (With cyclopentolate) revealed refractive error of -3.50 D Sphere/-3.75 D Cylinder at 174 degrees with spherical equivalent (S.E) of -5.50 D in right eye and -4.0 D sphere/-4.25 D Cylinder at 6 degrees with S.E of -6.25 D in left eye. Intracocular pressure and slit-lamp examination were within normal limits. There was no evidence of keratoconus in either eye on clinical examination and corneal topography. Detailed dilated fundus evaluation revealed normal optic disc and macula with a tessellated background with no evidence of pigmentary retinopathy.
CASE REPORT

Color vision and Electroretinogram (ERG) were within normal limits. A final diagnosis of Laurence-Moon-Biedl Syndrome with ametropic amblyopia due to compound myopic astigmatism was made and was advised best corrected glasses and occlusion therapy. The patient was explained the need for squint surgery at a later date.

The parents were counseled to bring the other two siblings for evaluation in the next visit. Interestingly, her two siblings aged 11 years (female) and 4 years (male) were found to have similar phenotypic features with postaxial polydactyly and central obesity (Figure 2, 3). The eldest sibling (BMI 24.7Kg/m2) (Case 2) also had mild intellectual disability. She had a BCVA of 6/18 and 6/24 in right and left eye respectively. Her cycloplegic auto refraction revealed a refractive error of -3.25 D Sphere/ -4.25 D Cylinder at 154 degrees (S.E -5.25D) in right eye and -3.25 D Sphere/-3.75D Cylinder at 9 degrees (S.E-4.5 D) in left eye. The youngest sibling (BMI 20.1Kg/m2) (Case 3) had moderate intellectual disability with small testes. His accurate BCVA could not be determined due to his intellectual disability. However, his cycloplegic auto refraction revealed a refractive error of -16.25 D Sphere/ -2.5 D Cylinder at 163 degrees in right eye (S.E-17.5) and -11.75 D Sphere/ -2.5 D Cylinder at 14 degrees in left eye (S.E-12). Both the siblings had a normal intra-ocular pressure with an unremarkable fundus and slit lamp examination. No abnormalities were detected on ERG and corneal topography. USG abdomen and pelvis of both the siblings were within normal limits. They too were diagnosed as Laurence–Moon–Biedl syndrome with ametropic amblyopia with compound myopic astigmatism and were advised best corrected spectacles for constant use.

At 3 months follow up, Case 1 showed a two-line improvement in visual acuity to 6/24 in both eyes with freely alternating divergent squint. Case 2 also showed improvement in her visual acuity of 6/12 in both eyes. Visual acuity in Case 3 could not be assessed properly because of his intellectual disability. Their social and behavioral attitude had also shown marked improvement because they were able to perform better in their school and communicate better with their peers.

DISCUSSION: Laurence-Moon-Biedl syndrome is an autosomal recessive condition characterized by rod-cone dystrophy, postaxial polydactyly, central obesity, mental retardation, hypogonadism, and renal dysfunction.1 The genes responsible for LMBS are located on chromosomes 16q21 (Type 2), 11q13 (Type 1), 3p12 (Type 3), and 15q22 (Type 4). The most common form of LMBS is type 1, and type 3 is the most rare form.2 According to the modified diagnostic criteria laid by Beales et al1, the diagnosis of Laurence Moon Biedl Syndrome is made if four of the primary features which include Rod-cone dystrophy, Polydactyly, Obesity, Learning disabilities, Hypogonadism in males, renal anomalies or three primary plus two secondary features which include Speech disorder/delay, Strabismus/cataracts/astigmatism, Brachydactyly/syndactyly, Developmental delay, Polyuria/polydipsia (Nephrogenic diabetes insipidus), Ataxia/poor coordination/imbalance Mild spasticity (Especially lower limbs), Diabetes mellitus, Dental crowding/ hypodontia/small roots/high arched palate, Left ventricular hypertrophy/congenital heart disease, Hepatic fibrosis. Our patients fulfilled four of the primary features except rod-cone dystrophy and one secondary feature i.e. high compound myopic astigmatism and hence were diagnosed as having this syndrome.
The common ocular findings in Laurence–Moon–Biedl syndrome include myopia, astigmatism, anisometropia, strabismus, keratoconus, pigmentary retinopathy, optic atrophy, nystagmus. Existing studies on Laurence–Moon–Biedl syndrome mention these ocular associations and the ERG patterns in this disease but none mention the pattern of refractive errors and their role as a potential amblyogenic factor in these patients.

Akinci et al. described the ocular and refractive findings in 17 patients with Laurence–Moon–Biedl syndrome. Of the patients evaluated, 88.2% had an ocular or refractive finding, 58.8% had myopia (degenerative in three cases), 52.9% had astigmatism, 17.6% had anisometropia, 11.7% had strabismus, 11.7% had retinitis pigmentosa, 5.9% had keratoconus, 5.9% had optic atrophy, and 5.9% had nystagmus. The pattern of the refractive errors and their association with strabismus has however not been elaborated upon in the above study.

ERG alterations have been detected in 52.6% cases with Laurence–Moon–Biedl syndrome. Histopathology and ERG point out to the uniform loss of the outer retinal layers, initially affecting rods but also rapidly affecting cones as the reason for the retinal dystrophy and night blindness in Laurence–Moon–Biedl syndrome. However, our patients did not have retinal dystrophy as evidenced by a normal ERG and absence of night blindness. Another possibility may be that the symptoms of night blindness could not be assessed properly due to intellectual disability or the signs of pigmentary retinopathy could be detected in their later age.

CONCLUSION: Uncorrected isolated high compound myopic astigmatism can be a cause of amblyopia and visual loss in Laurence–Moon–Biedl syndrome with the absence of night blindness and pigmentary retinopathy. The pattern of such high compound myopic astigmatism as noted in our study needs early detection and best corrective refraction to prevent visual loss. These patients need a yearly follow-up till their early teens for refractive correction and dilated fundus evaluation. Larger studies are however required to validate our findings.

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