There are some limitations to our study. Although we interviewed a heterogeneous group of strabismus patients, it is possible we missed some concerns that may be specific to a particular subpopulation of adults with strabismus, or may be present in other cultures. Nevertheless, we included each concern even if it was only mentioned once.

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Accommodative esotropia and Brown syndrome in a girl with recessive geleophysic dysplasia

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Geleophysic dysplasia and Weill-Marchesani syndrome are acromelic dysplasias characterized by short stature, brachydactyly, and joint contractures. Recessive Weill-Marchesani syndrome typically includes spherophakia, but the ocular phenotype of recessive geleophysic dysplasia is not well defined. We describe the ocular phenotype of a girl with genetically confirmed recessive geleophysic dysplasia (biallelic *ADAMTSL2* mutations). Features included high corneal astigmatism, accommodative esotropia, unilateral Brown syndrome, and no evidence for zonular disease at 12 years of age.

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Case Report

A 10-year-old girl previously diagnosed with familial geleophysic dysplasia and confirmed to harbor compound heterozygous mutations in the gene *ADAMTSL2* (c.338G>T/IVS5-89G>A) that segregated with the phenotype was referred for ophthalmic examination because of eye mislignment. The girl, born to first cousins, and her 2 affected sisters (not available for examination) were the subject of a prior report highlighting cardiac involvement in their disease.[1] Consistent with her diagnosis of geleophysic dysplasia, she had short stature, multiple infantile respiratory infections, joint contractures, and static cardiac valvular disease.[1] On examination, her affect and social interaction seemed appropriate for age. She was small (height, 115.4 cm [3rd percentile, 122 cm], weight 20.5 kg [3rd percentile, 19 kg]), with acromelia. Her facies were notable for thin upper lip, smooth and wide philtrum, anteverted nares, hypertelorism, and broad nasal bridge. She did not exhibit an abnormal head position. Visual acuity (without correction) was 20/60 in each eye. At near there was an esotropia of approximately 35^{Δ} and hypotropia of 10^{Δ}. Versions were significant for −2 elevation in adduction in the left eye that did not improve with ductions, consistent with Brown syndrome (Figure 1). Slit-lamp examination was normal, including no evidence for spherophakia or ectopia lentis before and after pharmacological pupillary dilation. Cycloplegic refraction (cyclopentolate 1%) was +5.50 −3.50 × 180 in the right eye and +6.00 −4.00 × 180 in the left eye. Fundus examination was significant for small crowded optic nerve heads in both eyes. She was prescribed her full hyperopic correction.

On follow-up examination 3 months later, visual acuity with correction was 20/20 in each eye. With correction, at near was an esotropia of 14^{Δ} and right hypotropia of 10^{Δ} in primary position. At 12 years of age her refraction and ocular motility were unchanged. Ocular biometry (Zeiss IOLmaster) was as follows: keratometry 42.99 D/48.08 D @072 in the right eye and 43.55 D/48.28 D @107 in the left eye; anterior chamber depth was 3.01 mm in the right eye and 3.06 in the left eye; and axial length was 20.92 mm in the right eye and 20.45 mm in the left eye.

Discussion

The ocular features of this child with recessive geleophysic dysplasia secondary to biallelic *ADAMTSL2* mutations were short eyes, high corneal astigmatism, and left Brown syndrome. There was no evidence for zonular disease at 12 years of age.

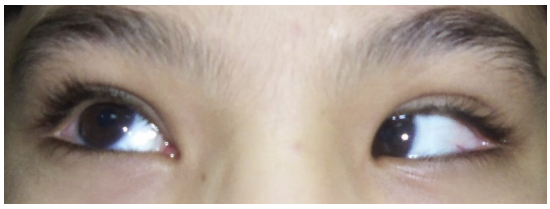

FIG. 1. During version up and to the right, the left eye has limited supraduction. This persisted with left duction up and to the right.

Both geleophysic dysplasia and Weill-Marchesani syndrome are acromelic dysplasias, which are characterized by short stature, brachydactyly, and joint contractures.[2] The recessive forms are caused by biallelic mutations in, respectively, the *ADAMTSL2* and *ADAMTS10* genes.[3,4] Findings more characteristic of geleophysic dysplasia include a round "happy" face (from *geleos*, meaning "happy" in Greek), recurrent middle ear and respiratory infections, cardiac valve anomalies, hepatomegaly, and tracheal stenosis.[2,3] Spherophakia with resultant progressive angle-closure glaucoma is a well-defined feature of Weill-Marchesani syndrome,[5] while most prior reports of genetically confirmed recessive geleophysic dysplasia either do not state whether there was ophthalmic phenotyping or simply mention the presence of "ophthalmic symptoms," with no further details.[3,6] There is one prior report of an adult diagnosed as geleophysic dysplasia for whom blue sclera, keratoglobus, and microspherophakia were mentioned as findings.[7] These ocular findings have never been reported in any other patient with geleophysic dysplasia. That patient also had recurrent intussusception, another finding which has not been reported in any other patient with geleophysic dysplasia. This raises the possibility that she may have had a different underlying diagnosis. That patient did have compound heterozygous *ADAMTLS2* variants, but both were intronic.

In the current genetically confirmed case, we specifically looked for and found no evidence for zonular weakness. As spherophakia is a consistent feature of Weill-Marchesani syndrome,[5] a lack of zonular weakness can be a useful feature to help distinguish geleophysic dysplasia from Weill-Marchesani syndrome. Although there are no data regarding age at which spherophakia develops in Weill-Marchesani syndrome, it is typically in early childhood.

Short eyes and resultant hyperopia were notable ophthalmic features in our case and are potentially additional useful distinguishing features from Weill-Marchesani syndrome. These features were also noted in a clinically diagnosed but not genetically confirmed 9-year-old girl who developed anterior chamber shallowing.[8] Hyperopia may be useful as a distinguishing feature from Weill-Marchesani syndrome, because Weil-Marchesani patients are typically myopic secondary to their spherophakia. We are unaware of any studies of axial length in Weil-Marchesani syndrome.

The high corneal astigmatism and Brown syndrome in our case could be coincidental or could be potentially be recurrent features of geleophysic dysplasia. Additional reports with detailed ophthalmic phenotyping of children with confirmed biallelic *ADAMTSL2* mutations are needed to definitively define the ocular phenotype for recessive geleophysic dysplasia.

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Infantile sialidosis: natural history in a preterm infant with two new pathogenic mutations and new ocular findings

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