OR *accessory* OR *anomalous* AND *extraocular muscle*; *Duane syndrome* AND *accessory* AND *extraocular muscle*; *accessory* AND *lateral rectus* AND *Duane syndrome*; *accessory* AND *lateral rectus* AND *type 2* OR *exotropic* AND *Duane syndrome*.

References

1. Duke-Elder KC, Wybar K. The Anatomy of the Visual system. System of Ophthalmology, vol 2. St. Louis, MO: Mosby; 1973: 427-8.
2. Khitri MR, Demer JL. Magnetic resonance imaging of tissues compatible with supernumerary extraocular muscles. Am J Ophthalmol 2010; 150:925-31.
3. Lueder GT. Anomalous orbital structures resulting in unusual strabismus. Surv Ophthalmol 2002;47:27-35.
4. Pineles SL, Velez FG. Accessory fibrotic lateral rectus muscles in exotropic Duane syndrome with severe retraction and upshoot. J AAPOS 2015;19:549-550.e1.
5. Molinari A, Plager D, Merino P, et al. Accessory extraocular muscle as a cause of restrictive strabismus. Strabismus 2016;24:178-83.
6. Knowlton PB, Mawn LA, Atkinson JB, Donahue SP. Strabismus resulting from an anomalous extraocular muscle in Gorlin syndrome. J AAPOS 2014;18:495-8.
7. Park CY, Oh SY. Accessory lateral rectus muscle in a patient with congenital third-nerve palsy. Am J Ophthalmolol 2003;136: 355-6.
8. Lueder GT, Dunbar JA, Soltal JB, Lee BC, McDermott M. Vertical strabismus resulting from an anomalous extraocular muscle. J AAPOS 1998;2:126-8.
9. Whitnall SE. An instance of the retractor bulbi muscle in man. J Anat Physiol 1911;46:36-40.

Ophthalmic diagnosis and optical coherence tomography of abetalipoproteinemia, a treatable form of pediatric retinal dystrophy

Arif O. Khan, MD,[a] Omar Basamh, MD,[b,c] and Hind M. Alkatan, MD[d]

A 16-year-old boy with early-childhood-onset retinal dystrophy and developmental delay was diagnosed with abetalipoproteinemia based on ophthalmic examination, history, and results of a peripheral blood smear. The diagnosis was confirmed by lipid profile and genetic testing, and an older sister was confirmed to be affected as well. Although abetalipoproteinemia is treatable in early childhood, most cases are diagnosed late if at all. We highlight clinical features that should raise suspicion for this treatable but likely under-diagnosed form of early-onset retinal dystrophy and document retinal optical coherence tomography findings for a genetically proven case.

Author affiliations: [a]Eye Institute, Cleveland Clinic Abu Dhabi, Abu Dhabi, United Arab Emirates; [b]Department of Ophthalmology, King Abdullah Medical City, Mecca, Saudi Arabia; [c]Division of Pediatric Ophthalmology, King Khaled Eye Specialist Hospital, Riyadh, Saudi Arabia; [d]Department of Ophthalmology, King Saud University, Riyadh, Saudi Arabia

Submitted August 11, 2018.
Revision accepted January 9, 2019.
Published online February 5, 2019.

Correspondence: Arif O. Khan, MD, Eye Institute Cleveland Clinic Abu Dhabi, PO Box 112412, Abu Dhabi, United Arab Emirates (email: arif.khan@mssm.edu).

J AAPOS 2019;23:176-177.

1091-8531/$36.00
https://doi.org/10.1016/j.jaapos.2019.01.005

Case Report

A 16-year-old boy was evaluated at King Khaled Eye Specialist Hospital for poor vision since early childhood. His 19-year-old sister was similarly affected, whereas his 3 eldest siblings (a brother, a sister, and a brother, in order of decreasing age) and a younger sister were asymptomatic. The boy's parents were from the same tribe. Poor vision had been noted since early childhood, particularly at night, and the parents were unsure whether it had been progressive. Review of systems revealed that both affected siblings had been "weaker" and delayed in their milestones compared to their siblings. Both affected siblings had not attended school because of perceived intellectual disability. Prior evaluation included brain magnetic resonance imaging, which was normal by report. Further questioning revealed that both siblings had problems with diarrhea after eating certain foods, particularly meat which they thus avoided. There was no history for polydactyly, labored breathing after birth, difficulty hearing, or cardiac disease.

The boy was under average for height and weight (153 cm and 45.60 kg). On ophthalmological examination, best-corrected visual acuity was 20/300 in each eye. There was pendular horizontal nystagmus and poor tracking. Anterior segment examination was normal. Retinal examination revealed a dystrophic retina, with frank central macular atrophy and overlying central gliosis. Optical coherence tomography revealed retinal thinning, particularly in the central macula. Autofluorescence showed an increased signal in the central macula. Electroretinography revealed cone-rod dysfunction.

Abetalipoproteinemia was suspected because of the atypical retinal dystrophy (with early central involvement) in the context of developmental delay and meat intolerance. A peripheral blood smear confirmed that the boy had marked acanthocytosis. Retinal findings and results of the blood smear are shown in Figure 1. Lipid profiles in millimoles/liter showed absent plasma triglyceride level (0.0 [normal, 5.2]) and decreased cholesterol profile (total cholesterol of 1.0 [normal 1.3]), with low-density lipoprotein (LDL) virtually absent (LDL of 0.1, HDL of 0.9). The affected sister had very similar ophthalmic findings and laboratory profile, and in addition to

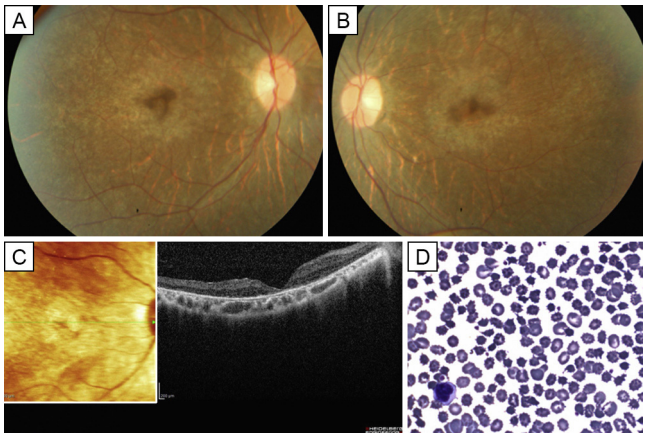

FIG 1. A-B, Fundus photographs showing a dystrophic appearance to the retina in both the right (A) and left (B) eyes, with choroidal show and central macular atrophy. C, Optical coherence tomography of the right eye showing outer retinal thinning, particularly in the central macula. The left eye was similar. D, Peripheral blood smear showing spiculated red cells, with surface projections of varying size (acanthocytosis).

acanthocytosis she also had anemia. Sanger sequencing of the gene *MTTP* (microsomal triglyceride transfer protein) confirmed that both siblings harbored a novel homozygous variant predicted to be pathogenic by several bioinformatic programs (c.1834G>C; p.Gly612Arg).

Discussion

Abetalipoproteinemia, also known as Bassen-Kornzweig syndrome,[1,2] is a rare syndromic form of retinal degeneration caused by complete absence of plasma apolipoprotein B–containing lipoproteins (chylomicrons, very low-density lipoprotein, and LDL).[3] These are carriers of fat and fat-soluble vitamins, and therefore with time affected children develop manifestations of deficient fat and fat-soluble vitamin absorption, including steatorrhea, muscle weakness, spinocerebellar ataxia, and retinal degeneration.[3] Marked acanthocystosis is typical and can sometimes lead to anemia. Biallelic mutations in the 97 kDa subunit of microsomal triglyceride transfer protein cause the disease.[4] The prevalence is estimated to be 1/1,000,000, but the condition is likely under-recognized. Mutations in the gene are often private to specific families or communities.[5]

Retinal degeneration is a prominent feature of abetalipoproteinemia, but most reports of the condition highlight extraocular features rather than the presentation to the ophthalmologist.[2,3] Children often present with central visual loss and an atypical retinopathy, although for some nyctalopia is the initial complaint. Yellowish dots and mottling are often seen in the posterior pole and midperiphery.[2] A gliosis can be appreciated over the central macula.[2] With time the entire retina is affected by pigmentary retinopathy. Angioid steaks have been reported,[6] as has helicoid peripapillary degeneration.[7] Because the condition is most amenable to treatment by vitamin supplementation at an early stage,[8] early diagnosis is important. In one study, none of the affected children who were treated before 2 years of age developed complications that are otherwise associated with the disease.[9] Early treatment can prevent or retard progression of retinopathy.[8] Unfortunately, systemic diagnosis and thus treatment are often delayed.

Most children with retinal dystrophy have ocular disease only. However, pediatric ophthalmologists should be aware that childhood-onset retinal dystrophy can be the sign of a systemic condition, particularly when in the setting of developmental delay. Neurometabolic disease is particularly important to identify, because in some cases it may be amenable to treatment when diagnosed early. Any child who presents with retinal dystrophy in the setting of developmental delay should undergo metabolic or genetic evaluation.

Our case highlights the ocular features of the treatable neurometabolic disease abetalipoproteinemia. The ophthalmologist can be instrumental in making the diagnosis. Confirmatory genetic testing or even a lipid profile may be difficult to obtain in some settings. However, a peripheral blood smear is an easily obtained and inexpensive test. We suggest that a peripheral blood smear to assess for acanthocytosis be considered when a child presents with retinal dystrophy in the setting of fatty food intolerance, muscle weakness, or neurological signs.

References

1. Bassen FA, Kornzweig AL. Malformation of the erythrocytes in a case of atypical retinitis pigmentosa. Blood 1950;5:381-7.
2. Kornzweig AL. Bassen-Kornzweig syndrome: present status. J Med Genet 1970;7:271-6.
3. Lee J, Hegele RA. Abetalipoproteinemia and homozygous hypobetalipoproteinemia: a framework for diagnosis and management. J Inherit Metab Dis 2014;37:333-9.
4. Shoulders CC, Brett DJ, Bayliss JD, et al. Abetalipoproteinemia is caused by defects of the gene encoding the 97 kDa subunit of a microsomal triglyceride transfer protein. Hum Mol Genet 1993;2:2109-16.
5. Stenson PD, Mort M, Ball EV, et al. The Human Gene Mutation Database: towards a comprehensive repository of inherited mutation data for medical research, genetic diagnosis and next-generation sequencing studies. Hum Genet 2017;136:665-77.
6. Gorin MB, Paul TO, Rader DJ. Angioid streaks associated with abetalipoproteinemia. Ophthalmic Genet 1994;15:151-9.
7. Wong AM, Héon E. Helicoid peripapillary chorioretinal degeneration in abetalipoproteinemia. Arch Ophthalmol 1998;116:250-51.
8. Chowers I, Banin E, Merin S, Cooper M, Granot E. Long-term assessment of combined vitamin A and E treatment for the prevention of retinal degeneration in abetalipoproteinaemia and hypobetalipoproteinaemia patients. Eye (Lond) 2001;15(Pt 4):525-30.
9. Runge P, Muller DP, McAllister J, Calver D, Lloyd JK, Taylor D. Oral vitamin E supplements can prevent the retinopathy of abetalipoproteinaemia. Br J Ophthalmol 1986;70:166-73.