A 5-month-old male infant was evaluated for visual inattention since infancy and intermittent nystagmus. Early hearing screening testing was failed. At 3 months of age, staring spells suggested seizures and were followed by normal electroencephalogram results; however, the neurologic examination suggested generalized hypotonia. The initial genetics evaluation focused on ocular hypertelorism and normal results for Noonan syndrome were reported. On examination, his cranium appeared large but measured in the third percentile (41 cm) associated with a large soft anterior fontanelle and ocular hypertelorism in the 95th percentile (49 cm). Bilateral simian creases were noted and an additional history was given of normal amniocentesis results at 21 weeks done for 21 trisomy and Smith-Lemli-Opitz syndrome. On further ocular examination, intermittent vague following was associated with visual inattention and no avoidance of a bright light. Cycloplegic refraction revealed no defects. Intermittent esotropia and up-jerk nystagmus were seen. The corneas were clear and intraocular pressures measured 20 mm Hg in both eyes. The pupils constricted to light but response seemed incomplete and slow. The irides were unpigmented but appeared dark in the periphery. Prominent thick strands crossed the pupils from the iris collarette circumferentially and contacted the anterior capsule centrally in both eyes. Dilated funduscopy allowed a good view. The discs were small and poorly vascularized. The choroid appeared hypoplastic with only moderate vascularization, giving the fundus a pale appearance. In the equatorial regions, multiple small, round, dark pigment spots were present (Figure A). Based on these ocular observations and the suspected retinal pathology characterized by loss of photoreceptor outer segments (Figure B), the clinical diagnosis was suspected. Serum testing for elevated long chained fatty acids and other metabolic indices of peroxisomal function was recommended and yielded positive results, and later genetic testing confirmed a compound heterozygote abnormality of the PEX1 gene.

What’s Your Diagnosis?
For the correct answer, see page 26
The correct answer to *What's Your Diagnosis?* is Zellweger syndrome spectrum disorder.

**REFERENCES**