A 37-year-old woman, gravida 1 para 0, at 29 weeks’ gestation, underwent amniocentesis due to a suspected polycystic kidney detected on routine fetal ultrasonography. While no genetic abnormality associated with polycystic kidney was identified (whole-exome sequencing Invitae), an incidental finding of a heterozygous c.501-2 ARB1 gene was noted. This specific RB1 SV had not been previously reported in any genetic databases, but was deemed pathogenic by Invitae, the commercial laboratory, in line with the American College of Medical Genetics and their guidelines for interpretation of sequence variants. No intraocular tumors were found on fetal sonography. There was no discernable family history of retinoblastoma or ocular disorders, and complete ophthalmic evaluation of both parents was normal with no suspected retinomas.

WHAT WOULD YOU DO NEXT?

A. Plan for early delivery

B. Continue routine prenatal follow-up

C. Further genetic evaluation of the fetus and parents

D. Advise pregnancy termination