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CASE REPORT

Anophthalmia Caused by Familial Homozygous Mutation of *VSX2*: a Case Report

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SUMMARY

Background: **Although rare, several mutations in the gene *VSX2* (visual system homeobox 2, formerly *CHX10*) have been associated with congenital autosomal recessive anophthalmia (absence of one or both eyes). This report describes a proband, who at presentation was gravida 2, para 0, and 30 weeks pregnant.**
Methods: **A 30-year-old woman with congenital bilateral anophthalmia was 30 weeks pregnant at the time of presentation. Her parents were fourth-generation collateral blood relatives, and the familial congenital disease history suggested a possible genetic cause for her anophthalmia. Next generation sequencing and Sanger sequencing of blood samples of the patient, her parents, and her husband were conducted. The fetus was examined via ultrasound.**
Results: **The woman patient had a homozygous variation of the *VSX2* gene (NM_182894.2) c.634delC (p.R211Gfs*90). Both of her parents carried a heterozygous variation of this locus. The husband showed no pathogenic variation in *VSX2*. The fetal ultrasound revealed bilateral eyeball lenses. A healthy girl was delivered at 41 weeks gestation, with bilateral eyeballs visible.**
Conclusions: **Homogenous mutation of *VSX2* c.634delC (p.R211Gfs*90) has not been reported previously. The patient's congenital bilateral anophthalmia was due to this homogenous mutation, the result of familial inbreeding. Avoiding near-relative marriage is an important means of preventing such diseases.**
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Supplementary Tables and Figures

Figure 1. The patient's binocular picture, displaying the absence of the eyeballs.

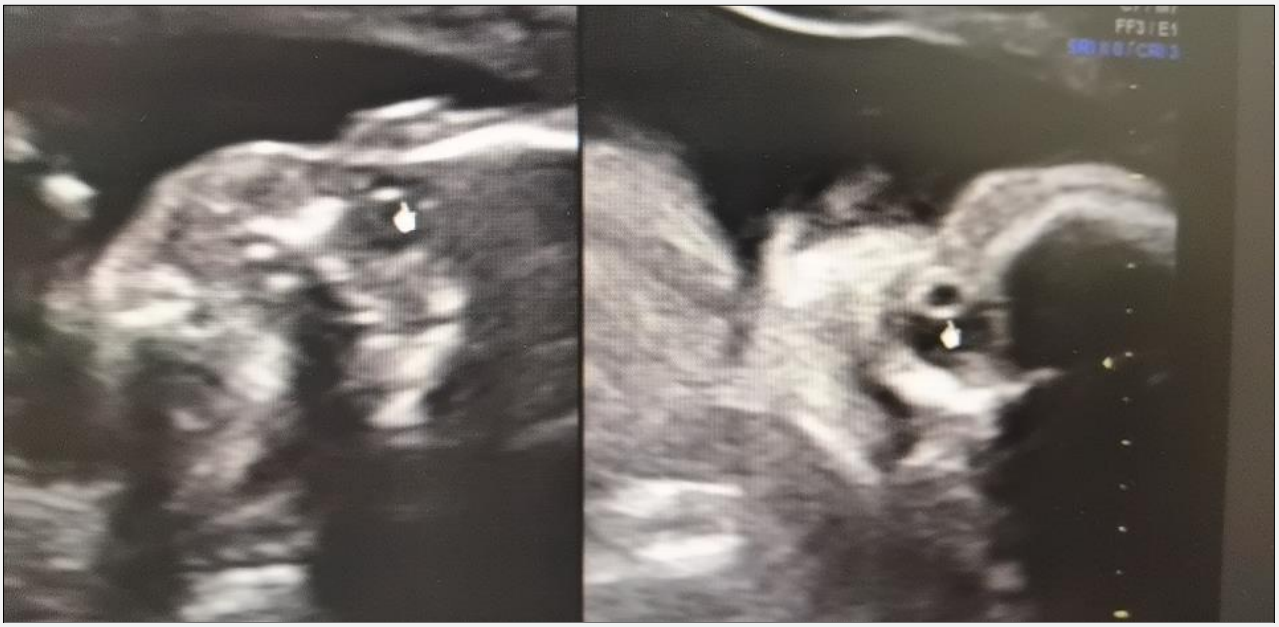

Figure 2. Fetal ultrasound with visible bilateral eyeball lenses (hyperechoic film shown in the icon).