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Research Letter

Congenital cataracts diagnosed by prenatal ultrasound

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Most cataracts are related to aging. By contrast, congenital cataracts are rarely detected at birth. Moreover, prenatal diagnosis of congenital cataracts is rare. Congenital cataracts occur in one to six newborns out of 10,000 newborns [1–3] and may be associated with congenital infections, chromosomal or genetic defects, or multiple anomalies and syndromes. We present here an interesting case of congenital cataracts diagnosed by prenatal ultrasound (US).

A 25-year-old pregnant woman, gravida 1, para 0, was healthy before pregnancy. Her prenatal care was carried out at a local clinic and was uneventful. Amniocentesis was not performed. She was referred to our center at 22 weeks of gestation due to a suspected increased nuchal thickness. Level II US revealed increased nuchal thickness (0.6 cm) and left pyelectasis (5 mm). Notably, bilateral extremely dense echogenic lenses—suspected congenital cataracts—were depicted remarkably by two-dimensional (2D) US (Figure 1). On examination with three-dimensional (3D) US, the diagnosis of congenital cataracts was strongly confirmed with a significantly dense echogenic structure in the fetal lens (Figure 2). The pregnancy was then terminated after genetic consultation with the couple. Chromosome analysis from cord blood revealed a 46, XY normal male karyotype. Grossly, no anomaly was found in this fetus, except congenital cataracts. A toxoplasmosis, rubella, cytomegalovirus, herpes simplex, and human immunodeficiency virus profile in fetal blood showed no evidence of recent infection. Autopsy revealed vacuole formation and necrosis of bilateral lenses compatible with congenital cataracts (Figure 3). Mild left renal pyelectasis was also noted. The cause of congenital cataracts in this case remained idiopathic.

Second trimester US can screen fetal lens anatomy, which is an anechoic structure under normal conditions and presents as a hollow space in 3D images (Figure 2A). As for a cataractous lens, it is

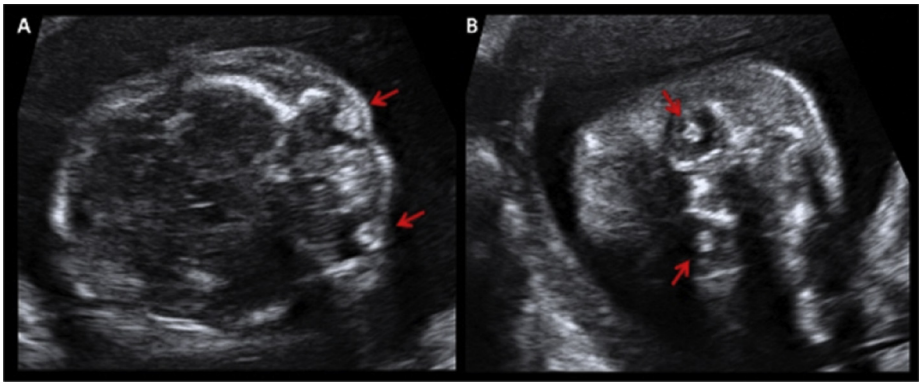

Figure 1. Two-dimensional prenatal ultrasound images. Bilateral dense echogenic lenses are indicated by arrows. (A) Transverse view. (B) Coronal view.

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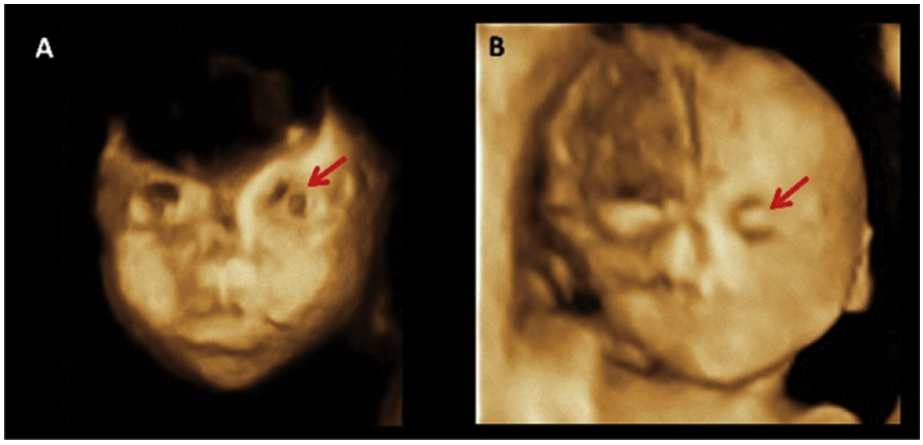

Figure 2. Three-dimensional (3D) reconstructed images of the second trimester lens. (A) Normal lens is anechoic and presents as a hollow space in a 3D image (red arrow). (B) Cataractous lenses are dense echogenic structures (red arrow).

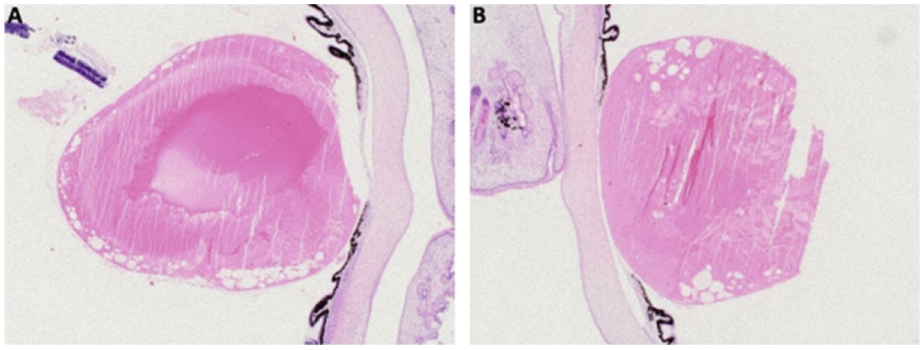

Figure 3. Images from autopsy of bilateral lenses. Vacuole formation and necrosis of the (A) right lens (20×) and (B) left lens (20×) are shown.

densely echogenic, unilateral or bilateral, and presents as dense echogenic structure in 3D images. Three-dimensional US could be applied for assessment of cataracts. To our knowledge, there are few literature reports of congenital cataracts diagnosed by prenatal 3D US [4]. Our case showed the remarkable 2D and 3D pictures and confirmed the findings by pathology.

Family history is an important clue for prenatal diagnosis of congenital cataracts. Thus thorough history taking is important during prenatal examination. In our case, there was no associated family history. But in other cases, congenital cataracts may be hereditary and are associated with genetic defects. The defect could be inherited in one of the three patterns: autosomal dominant, autosomal recessive, or X-linked transmission. Among which, autosomal dominant inheritance is still the most frequent, with almost complete penetrance but variable expressivity [5]. Chromosomal defects, such as trisomy 21, 13, and 18, are also related to congenital cataracts. Medical diseases such as galactosemia, hypoparathyroidism, and maternal diabetes are also related to congenital cataracts and should be excluded by laboratory examination. Intrauterine infections (toxoplasmosis, rubella [6], cytomegalovirus, herpes simplex virus, varicella [7], and syphilis) are also common causes of cataracts. In developing countries, congenital cataracts are more common, probably due to a high prevalence of congenital infection. The outcome for fetuses with congenital cataracts depends on the associated syndrome. Once congenital cataracts are noted, other possible fetal anomalies should be searched for systemically and carefully.

Conflicts of interest

The authors have no conflicts of interest relevant to this article.

References

[1] Abrahamsson M, Magnusson G, Sjöström A, Popovic Z, Sjöstrand J. The occurrence of congenital cataract in western Sweden. Acta Ophthalmol Scand 1999;77:578–80.
[2] Rahi JS, Dezateux C. British Congenital Cataract Interest Group. Measuring and interpreting the incidence of congenital ocular anomalies: lessons from a national study of congenital cataract in the UK. Invest Ophthalmol Vis Sci 2001;42:1444–8.
[3] Francis PJ, Berry V, Bhattacharya SS, Moore AT. The genetics of childhood cataract. J Med Genet 2000;37:481–8.
[4] Pooh RK, Kurjak A. Recent advances in 3D assessment of various fetal anomalies. DSJUOG 2009;3:1–23.
[5] Deng H, Yuan L. Molecular genetics of congenital nuclear cataract. Eur J Med Genet 2014;57:113–22.
[6] Wolff SM. The ocular manifestations of congenital rubella. Trans Am Ophthalmol Soc 1972;70:577–614.
[7] Lambert SR, Taylor D, Kriss A, Holzel H, Heard S. Ocular manifestations of the congenital varicella syndrome. Arch Ophthalmol 1989;107:52–6.