Identical Choroid Plexus Cysts in Monozygotic Monochorionic Twins

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Abstract

- Choroid plexus cysts have been infrequently reported with chromosomal abnormalities.
- Isolated choroid plexus cysts in a monozygotic twin pair hints to a genetically determined pathway as a possible cause.

Choroid plexus cyst is detected in less than 1% of second trimester fetuses [1]. The natural history varies from spontaneous resolution by the third trimester [2] to progressive enlargement [3]. Large choroid plexus cysts may even result in obstructive hydrocephalus requiring external cerebrospinal fluid drainage.

Choroid plexus cysts have been infrequently reported with Down syndrome [4] and other chromosomal abnormalities [5]. However, whether the formation of cysts is related to these abnormalities is debated [2]. The controversy of genetically determined pathways is even higher in cases of isolated choroidal plexus cysts.

Congenital malformations have been investigated in genetically identical monozygotic twins in order to identify concordant malformations that are likely to have a genetic basis rather than occur secondary from isolated mutations [6]. Our observation of isolated choroid plexus cysts in a monozygotic twin pair hints to a genetically determined pathway as a possible cause.

Case Report

A 32-year-old woman underwent fetal ultrasound screening at 17 weeks of gestation and was found to have monochorionic monozygotic male twins. The first twin had severe fetal bradycardia and cardiomegaly (cardiothoracic diameter ratio of 2). The second twin appeared to have no cardiovascular defects. At cranial ultrasound assessment, both twins showed an isolated choroid plexus cyst on the right side. In the first twin, the cyst measured 3.7 mm × 7.1 mm (Figure 1A); in the second twin, the cyst measured 4.8 mm × 0.8 mm (Figure 1B). Fetal karyotyping was normal.

Discussion

The presence of identical isolated choroid plexus cysts in monochorionic monozygotic twins support the possibility that these central nervous system anomalies are genetically determined. Unlike other fetal abnormalities and congenital malformations [6], concordant patterns among monozygotic twin pairs have not been studied for choroid plexus cysts. In part, such lack of studies may be attributable to the rarity of the isolated cysts [1] and spontaneous obliteration in third trimester [2]. In both twins, other cephalic parameters were within normal range for gestational period.

Histologically, the choroid plexus cyst is composed of two layers; a fibrous outer membrane and an inner layer of cuboidal choroid plexus epithelium that may be ciliated cells [7,8]. There are two main components of the choroid plexus; the epithelium derived from the neural tube epithelium and mesenchyma derived from the meninges. The cyst formation is most likely due to aberrant growth of either neuroepithelium [7] or angiomaticus interconnecting thin-walled capillaries [9] within the matrix of immature choroid plexus. The aberrant growth is anticipated to occur during the ninth developmental week as the embryonal capillary net is replaced by elongated loops of wavy capillaries that lie under regular longitudinal epithelial cells [9]. Several pathways, such as Notch3 signaling, the transcription factor TWIST1, platelet-derived growth factor receptor, and the tumor necrosis factor-related apoptosis-inducing ligand pathways, are active within the choroid plexus [10]. It remains unclear whether the genetic patterns that lead to formation of choroid plexus cysts are affecting the cell lines or the expression of growth modifying pathways within the choroid plexus. The possibility that...
concordant choroid plexus cysts in twins may be related to common placental or amniotic fluid factors cannot be excluded.

The occurrence of identical isolated choroid plexus cysts in monozygotic twins would support the possibility of concordant alteration or modification in genotype of the cell lines involved in formation of choroid plexus.

**References**