Topic: Prenatal Diagnosis

Monozygotic twins discordant for trisomy 4

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Discordant karyotype of MZT twins are rare.

We present the first case reported to date of monochorionic diamniotic (MC-DA) twins with discordant karyotype manifested as mosaic trisomy 4 (twin A) and a normal karyotype (twin B).

A 32-year-old healthy woman in her first spontaneus pregnancy is described.

First trimester ultrasound revealed a MC-DA gestation with a large cystic higroma in twin A.

Discordant karyotype on amniocentesis were obtained: twin A revealed a trisomy 4 (47,XX,+4), whereas twin B was normal (46, XX).

The 20-week scan revealed in twin A a SUA, growth restriction, microcephaly, corpus callosum agenesis, ventriculomegaly, cerebellum hypoplasia, spina bifida, thumb anomalies, retrognatia, clubfeet, diaphragmatic hernia and left kidney agenesia. Twin B appeared structurally normal.

Selective twin feticide versus expectant management were offered. Parents chose the second option.

The woman underwent spontaneous labor at 33+5 weeks: twin A weighed 995 g (below 1st centile), Apgar score 1/1 and died 60 min after birth; normal live born twin B weighed 1,950 g. The postmortem study confirmed the sonographic findings in twin A and MC-DA placenta.

Twin A postnatal peripheral blood showed a 46,XX karyotype and cultured fibroblast showed 47, XX, +4 karyotype. Twin B showed normal karyotype.

Molecular genetic analysis by QF-PCR assay with polymorphic markers confirmed monozygocity.

Trisomy 4 mosaicism has not been reported in any of the large studies on CVS mosaicism and only rarely on amniocentesis. Only four cases have been reported to date. This is the first reported on discordant MC-DA twins.

Common phenotype of trisomy 4 mosaicism includes most features found in our case.

As the degree of mosaicism is often related to the severity of clinical phenotype, high degree of mosaicism is suspected in our case.

Trisomy 4 mosaicism may be another example of tissue-limited mosaicism as the case reported here.