ABSTRACT

Short rib polydactyly syndrome (SRPS) is a rare inherited, lethal skeletal dysplasia, characterized by a triad of micromelia, polydactyly, and short horizontal ribs. We report a case of SRPS type 2, also called Majewsky syndrome, the rarest entity that was diagnosed by antenatal sonographic features at 21 weeks of gestation.

CASE REPORT

A 27-year-old woman (G1P0) without consanguinity or remarkable medical records was referred to our maternal hospital at 20-21 weeks of gestation. First trimester ultrasound screening showed an NT measurement of 1.5mm, however, combined screening test was not performed. The level-two ultrasound examination at 21 weeks of gestation revealed an extremely narrow thorax and short ribs. The upper and lower limb bones were found shorter than expected values (-3SD) associated with bilateral polydactyly. A combination of these signs strongly evoked a SRPS. Additionally, other non-skeletal manifestations were demonstrated, including Dandy Walker malformation, critical aortic stenosis, mitral valve stenosis and ventricular disproportion. In the presence of severe pathologic features, and following a prenatal counseling, the couple opted for termination of. However, they denied fetal autopsy or any molecular genetic analyses.

Clinically postmortem examination confirmed prenatal ultrasound findings of a SRPS, including bilateral polydactyly in both pre- and postaxial pattern. Syndactyly was not seen on antenatal assessment. The facial dysmorphism was finally revealed, showing broad forehead, hypertelorism, low-set ears, flattened nasal bridge, median cleft lip and soft palate.

These findings were in favor of a SRPS type 2 as previously described in the literature.

CONCLUSION

Short rib polydactyly syndrome (SRPS) can be diagnosed prenatally by a constellation of sonographic characteristic features. However, molecular genetic analysis and postmortem phenotypic description are indispensable for accurate counseling since this condition can recur as an autosomal recessive trait.