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Short Limbs, Earlier Diagnosis: Antenatal Presentation of Cornelia de Lange Syndrome

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CASE

- 30-year old female presented at 35 weeks gestation to Maternal-Fetal Medicine with intrauterine growth restriction (IUGR)
- 9 weeks: Demise of two fetuses in the triplet trichorionic gestation
- 11 weeks: Growth restriction of the remaining triplet noted

ULTRASOUND RESULTS

- Micromelia with long bones measuring <2 SD below the mean
- Femur:foot length <1
- Frontal bossing
- Small chest circumference
- Small ventricular-septal defect (VSD)

MRI RESULTS

- 54% observed:expected fetal lung capacity

MANAGEMENT of PREGNANCY

- Betamethasone for fetal lung maturity
- Cesarean section at 38 weeks 6 days gestation

MANAGEMENT of NEONATE

- Post-delivery microarray demonstrated normal female chromosomes
- NICU admission, intubated due to respiratory distress syndrome
- Severe gastric reflux warranted gastric tube placement
- Neonatal echo consistent with small VSD
- 2 months old: Diagnosis of Cornelia de Lange Syndrome (CdLS) made at skeletal dysplasia clinic

CONCLUSION

- Negative genetic testing does not exclude the diagnosis of CdLS, which should remain in the differential diagnosis with antenatal findings of frontal bossing, micromelia, and first trimester IUGR.

CLINICAL SIGNIFICANCE

- The incidence of CdLS is estimated to be 1:10,000-30,000 newborns.
- Most cases result from new mutations in the HDAC8 or SMC1A gene with no known family history
- One third of all cases have unknown etiology
- Many cases are diagnosed at birth or after birth, as this patient was diagnosed at 2 months old.

- The findings of IUGR extremely early in the pregnancy (usually noted in the 3rd trimester, not the 1st trimester) and detailed ultrasound revealing micromelia and frontal bossing are signs that could lead providers to include CdLS on the differential diagnosis during the antenatal period.
- With these antenatal findings, postnatal genetic evaluation may be warranted despite a normal antenatal genetic workup.

POINTS FOR DISCUSSION

- Are antenatal ultrasound findings reliable for a clinical diagnosis of CdLS?
- Would an earlier diagnosis of Cornelia de Lange Syndrome affect management and outcome?