Objectives: We present different fetal brain anomalies manifesting with a kinked brainstem (KB).

Methods: 5 fetuses with a KB were evaluated in 2nd trimester.

Results: Associated CNS abnormalities included: ventriculomegaly (VM) (4 cases), complete or partial agenesis of corpus callosum (ACC) (4), abnormal or delayed cortical development (4), vermian hypoplasia (VH) (4).

Fetus 1 (19 GW): KB, mild VM, absent Sylvian fissures, partial ACC, dysmorphic and enlarged tectum, and a small upward rotated vermis, diagnosed as having cobblestone malformation confirmed by autopsy.

Fetus 2 (19 GW): KB, underdeveloped cerebral hemispheres, mild VM, complete ACC, thin pons, and VH, presumed to have a tubulinopathy. Autopsy confirmed the prenatal findings, cortical histology was normal for gestational age.

Fetus 3 (15 GW): KB, a thick nuchal fold, pelvic kidney, severe VM and absent CSP, male gender, presumed to have L1 syndrome. The autopsy (22 GW) revealed complete ACC, KB, hypoplastic cortico-spinal tracts, and fragmented dentate nuclei. Cortical histology was normal for gestational age.

Fetus 4 (17 GW): KB, occipital encephalocele, polycystic kidneys, and postaxial polydactyly, diagnosed as having Meckel-Gruber syndrome. This was the second pregnancy of consanguineous parents, the previous one was terminated for identical malformations.

Fetus 5 (19 GW): KB, alobar holoprosencephaly with a dorsal cyst, a small dysplastic vermis associated with cystic dilatation of the fourth ventricle.

Conclusion

Brainstem kinking has been reported mainly in α-dystroglycanopathies. Our cases indicate that a KB can be associated with a spectrum of severe CNS malformations, being the earliest manifestation in some of them.