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Clinicopathological characteristics of type III lissencephaly: A case report

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Type III lissencephaly is reported as a lethal familial disorder (OMIM 601160). It is a syndromic entity that comprises intrauterine growth retardation (IUGR), fetal akinesia deformation sequence (FADS), severe microcephaly, agyria, spino-pontocerebellar atrophy and diffuse neurodegeneration. We report a 22-week male fetus who achieved typical features of type III lissencephaly, which led to termination of pregnancy. Ultrasonography showed IUGR, polyhydramnios, microcephaly, hydrocephaly, cerebellar hypoplasia and limb deformities. The fetus was born to nonconsanguinous parents with a healthy child. Array CGH didn't detect genomic imbalances. Detailed fetopathological examination was carried out after parental consent. It confirmed the IUGR and revealed multiple anomalies including craniofacial dysmorphism, short and wide neck, FADS, genital hypoplasia, severe four-limb amyoplasia, thoracic scoliosis, pulmonary hypoplasia, cardiac hypotrophy, adrenal hypoplasia, common mesentery and single umbilical artery. The neuropathological examination showed external hydrocephalus with enlarged subarachnoid spaces, hydranencephaly, severe spino-pontocerebellar hypoplasia and corpus callosum agenesis. Histology revealed a decrease of the testicular Leydig cells and confirmed the clinical impression of neurogenic muscular atrophy. It disclosed a diffuse neurodegeneration involving the grey and white matter as well as atrophy of the corticospinal tracts and vermian agenesis. Additionally, degeneration of motor neurons in the spinal anterior horns was noted. Microcalcifications were found in the cerebral hemispheres and brainstem. In conclusion, the type III lissencephaly is a central nervous system neurodegenerative disease with early prenatal onset. It's a very challenging condition because its molecular basis is not yet been elucidated. Thus, an adequate genetic counselling cannot be given to the parents.

Biography

Sihem Darouich, MD, is a Fetopathologist at the University Hospital Habib Bougatfa of Bizerte and Professor at the Department of Histology and Embryology, Faculty of Medicine of Tunis, Tunisia. Over the last several years, she has focused on fetal genetic syndromes, especially syndromic multicystic renal dysplasia and skeletal dysplasias. She is a member of the French Society of Fetopathology (SoFFoet) and of an editorial board of a reputed international journal.

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