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The usefulness of fetopathological examination in the diagnosis of Blomstrand chondrodysplasia: Report of an autopsy case

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Blomstrand chondrodysplasia is a very rare lethal skeletal dysplasia due to loss-of-function mutations in the gene encoding the type 1 parathyroid hormone receptor (PTHr). It is associated with resistance to the PTH that results in acceleration of endochondral ossification and bone remodeling deficiency. Our aim is to discuss the clinico-pathological characteristics of Blomstrand chondrodysplasia in a 21-week male fetus and the usefulness of the fetopathological examination in the diagnosis of this condition. Antenatal ultrasound showed severe tetramicromelia and hydrops fetalis, leading to the termination of pregnancy. Postmortem radiographs showed increased bone density, accelerated bone maturation (presence of points of ossification in pubis and calcaneum), irregular short ribs, short tubular bones with wide metaphyses and ossification of the hyoid bone and laryngeal cartilages. Macroscopic examination confirmed the fetal hydrops and the extreme tetramicromelia, and demonstrated characteristic craniofacial dysmorphism including mild exophthalmos, hypoplastic nose, protrusion of the tongue, micrognathia, low-set and badly-hemmed ears. Besides, short and wide neck, markedly narrow thorax, prominent abdomen, broad and short hands and feet, and pulmonary hypoplasia were also noted. On microscopy, the hyaline cartilage exhibited a compact and dense appearance and ectopic points of ossification without obvious primary ossification center. Additionally, the growth plate was often completely absent with presence of few columns of hypertrophic chondrocytes, lack of a readily identifiable proliferative zone, very irregular ossification line, thick layers of endochondral bone matrix and decreased size of the marrow spaces.

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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