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Usefulness of Fetopathological Examination in the Diagnosis of Skeletal Dysplasias

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The skeletal dysplasias (SD) are the most common of the fetal genetic skeletal disorders. Classification of these conditions is based on clinical, radiological and pathological criteria [1]. The identification of an increasing number of genes involved in these disorders led to consider a molecular classification that groups phenotypically different conditions caused by mutations in the same gene [2]. Thus, the SD are composed of distinct groups of squeletal disorders covering a wide spectrum of gravity. However, this classification is still hybrid because the molecular defects have not yet been identified in all cases of SD [2]. Prenatal imaging can detect early fetal skeletal anomalies of varied severity and prognosis. It can accurately predicts lethality, based on the identification of pulmonary hypoplasia and severe limb shortening, but often raises a prenatal diagnostic dilemma [3]. Similarly, a targeted molecular study is difficult in a situation of fortuitous prenatal detection of a SD. Thus, fetopathological examination after pregnancy interruption is important to provide an accurate diagnosis [1].

Group/Name of Disorder	Skeletal dysplasia	Number of cases (%)
Osteogenesis imperfecta and decreased bone density group (group 25)	Osteogenesis imperfecta, perinatal lethal, severe and moderate forms	21 (29)
FGFR3 chondrodysplasia group (group 1)	Thanatophoric dysplasia type 1 (TD1) and type 2 (TD2)	18 (25)
Ciliopathies with major skeletal involvement group 9	Chondroectodermal dysplasia (Ellis-van Creveld) Short rib-polydactyly syndrome (SRPS) type 3 (Verma-Naumoff) Asphyxiating thoracic dysplasia (ATD; Jeune) Short rib-polydactyly syndrome (SRPS) type 2 (Majewski)	9 (12)
Sulphation disorders group (group 4)	Achondrogenesis type 1B (ACG1B) Diastrophic dysplasia (DTD)	7 (10)
Abnormal mineralization group (group 26)	Hypophosphatasia, perinatal lethal form	3 (4)
Type 2 collagen group (group 2)	Hypochondrogenesis Kniest dysplasia	2 (3)
Perlecan group (group 5)	Dyssegmental dysplasia, Silverman-Handmaker type	2 (3)
Filamin group and related disorders (group 7)	Osteodysplasty Melnick-Needles Otopalatodigital syndrome type 2	2 (3)
Severe spondylodysplastic dysplasias (group 14)	Achondrogenesis type 1A (ACG1A)	2 (3)
Campomelic dysplasia and related disorders (group 18)	Campomelic dysplasia (CD) Stûve-Wiedemann dysplasia	2 (3)
Chondrodysplasia punctata (CDP) group (group 21)	CDP, X-linked dominant, Conradi-Hûnermann type (CDPX2) Greenberg dysplasia	2 (3)
Mesomelic and rhizo-mesomelic dysplasias (group 17)	Langer type (homozygous dyschondrosteosis)	1 (1)
Neonatal osteosclerotic dysplasias (group 22)	Blomstrand dysplasia	1 (1)

Table 1: Prevalence and pattern of skeletal dysplasias.

Here, taking into account the classification of genetic skeletal disorders, 2015 revision (Nosology 2015), we chose to classify the SD according to their most striking phenotypical and radiological criteria regardless of lethality. In a combined retrospective and prospective study, we have gathered radiological, macroscopic and histological data on fetuses diagnosed with SD following abortion, stillbirth and immediate postnatal death over an 8-years period (2009-2016) at the Department of Embryo-Fetopathology of Tunis. Of the 5995 fetal autopsies carried out, 72 (1.2%) were diagnosed with SD. The prevalence and pattern of SD in fetopathological practice are reported in Table 1.

These 72 cases encompassed 13 of 42 groups of Nosology 2015. The more common Nosology groups of SD were osteogenesis imperfecta (29%), fibroblast growth factor receptor type 3 (FGFR3) chondrodysplasias (25%), ciliopathies with major skeletal involvement (12%) and sulphation disorders (10%). Fetal age at autopsy ranged from 12 to 40 weeks with a mean of 22 weeks. There was a slight predilection for male gender among SD (male to female ratio: 1.15).

In conclusion, we have mentioned the commonly observed SD in fetopathology without being able to be exhaustive. Thus, our brief comment points to the great diversity and complexity of these disorders. Their enumeration and comprehension are far from being complete. Therefore, fetopathological examination is of extremely great importance as it allows to give an accurate diagnosis, guide the genetic study and provide genetic counseling.

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