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Skeletal dysplasia charts

SKELETAL DYSPLASIAS PRESENTING AT BIRTH

DIAGNOSIS †	ALWAYS LETHAL *	LIMBS					SKULL	RIBS		SPINE		PELVIS	BONE DENSITY ▲ ▼
		SHORT	BENT/ ANGULATED	DISLOCATION	FEMUR/ TIBIA / FIBULA	POLY-DACTYLY	OSSIFICATION	SHORT	BEADED/ FRACTURED	PLATY-SPONDYLY	SOME ABSENT OSSIFICATION	"TRIDENT" "SPIKEY"	+ FRACTURE
ACHONDROPLASIA - HETERO:		rhizo.						+				+	
- HOMO:	+	rhizo.						+				+	
THANATOPHORIC I	+	++	+					++		++		+	
II	+	+	+				Clover leaf	+		+		+	
(Thanatophoric Variants)													
ACHONDROGENESIS I	+	++					▼	+	+		+		
II	+	++						+			+		
HYPOCHONDROGENESIS	+	+						+		+	+		
SPONDYLO-EPIPHYSEAL DYSPLASIA CONGENITA		(rhizo.)						+		+			
OSTEOGENESIS IMPERFECTA IIA	+	+	+				▼		+	+			▼ +
IIB	+	+	+						+				▼ +
IIC	+	+	+				▼		+				▼ +
III		(+)	(+)						+				▼ +
HYPOPHOSPHATASIA Lethal	+	+			Fib. ▼		▼		+	+	+		▼ +
Surviving			+										
CAMPOMELIC	+		+	+	Fib. ▼								
KYPHOMELIC		rhizo.	+										
DIASTROPHIC		+ (1st M.C. ++)		+						(+) (Scoliosis)			
METATROPIC		+			Dumb-bell			+		++			
KNIEST		+			Dumb-bell					(+)			
FIBROCHONDROGENESIS	+	+			Dumb-bell			++		++			
CHONDRODYSPLASIA PUNCTATA		rhizo. (+)											stippled ectopic calcification
OSTEOPETROSIS							▲						▲ +
PYCNODYSOSTOSIS							▲						▲ +
I Saldino-Noonan	+	+				+		++				+	
SHORT II Majewski	+	meso.			Oval Tibia	+		++					
RIB III Verma-Naumoff	+	+				+		++				+	
SYNDROMES Beemer	+	meso.				(+)		++					
Asphyxiating Thoracic (Jeune)		(+)				(+)		++				+	
Ellis van Creveld		meso.				+		++				+	
Atelosteogenesis I	+	rhizo.	+	+	Fib. ▼			+		+			
II	+	rhizo.		+	Fib. ▼								
III		rhizo.		+	(Fib. ▼)								
Boomerang / De La Chappelle	+	+	+		Fib. ▼			+		+	(+)	(+)	
Opsismodysplasia		+						+		+	+	+	
Schneckenbecken	+	+			Dumb-bell			+			+	+	
Dyssegmental Dysplasia I		+	+					+					
II	+	+	+					+					

All these findings should be easily observed by the non-specialist

† Grouped according to main radiographic features and (approximately) in descending order of frequency.

* Short limbs can be identified on ultrasound by 20 weeks gestation in the lethal conditions and a few non-lethal.

( ) In parenthesis – the finding may be present but mild, or absent.

▼ – indicates: 'reduced, short, hypoplastic or absent'.

▲ – indicates: 'increased, dense'.

Tissues should be stored from all these severe skeletal dysplasias to enable confirmatory genetic testing and/or prenatal diagnosis. Cryopreserved fibroblasts are suggested.

An approach to the radiological diagnosis of lethal and other skeletal dysplasias presenting at birth. (A chart prepared by R. Wynne-Davies C.M. Hall and J. Hurst for the Skeletal Dysplasia Group for Teaching and Research, Occasional publication no. 8c (2004).)