



THE NON-LETHAL

DIAGNOSTIC GROUPS †	X RAY EVIDENCE + UNDER 2yrs ▼ 2yr OVER ▲	STATURE and BODY PROPORTIONS				SKULL				SPINE				
		INCREASED	REDUCED			CRANIUM		FACIAL BONES	MANDIBLE	Scoliosis/Kyphosis	Persistent Ovoid	Pleuroplegomy	Odontoid Dysplasia	
			Trunk	Limbs	Rhizo:	Mesomelic	Size							Density
							Suture Fusion Early ▲ Late ▼ Wormian W							
I	MULTIPLE EPIPHYS: DYS: HERED: ARTHRO: OPHTH: CHONDRO: PUNCTATA (Severe & mild forms) (Conradi)	▲ ▲ ▼ ▼ ▼ ▲		(+)				▼ (▼)	(▼)	(+) +		(T11, 12) +		
II	SPONDYLO-EPIPHYS: Congenita Tarda - X Linked - Dom: Rec. - Progressive arth:	▼ ▲ ▲ ▲ ▲ ▲ ▲ ▲		+ + + + (+)				(▼)		(+) + + +		+ + + +		
III	METAPHYS: DYS: - SCHMID - McKUSICK - JANSEN - with MALABSORP., NEUTROPEN: SPONDYLO-METAPHYSEAL DYS:	▲ ▲ (▼) ▲ ▼ ▲ ▲ ▲		Lower + + +		▼ ▲	(▲) (▲)	+ +			(+) (+)		(+) (+)	
IV	ACHONDROPLASIA HYPOCHONDROPLASIA DYSCHONDROTEOSIS ACROMESOMELIA ACRODYSOSTOSIS	▼ ▲ ▲ ▲ ▲ ▲ ▼ ▲ ▲ ▲		+ + + + + + + + (+)		▲ (▲)		(▼) (▼)		T-L Kyphos				
V	PSEUDOACHONDROPLASIA DIASTROPHIC DYSPLASIA METATROPIC DYSPLASIA KNIEST DISEASE DYGGVE-MELCHIOR CLAUSEN CHONDRO: ECTO: (Ellis van Creveld) ASPHYX: THOR: DYS (Jeune)	▲ ▲ ▼ ▲ ▼ ▲ ▼ ▲ ▲ ▲ ▼ ▲ ▼ ▲		+ + + + + + + + + + (+) + (+) +									(+) (Triangular) (+) + ++ + (+) + + + Teeth ▼	
VIII	STORAGE DISEASES MPS - Hurler - Hunter - Scheie - Sanfilippo - Mannoosidosis - Maroteaux-Lamy - Morquio MLS - GM, Gangliosidosis Type 1	▼ ▲ ▲ ▲ ▲ ▲ ▲ ▲ ▲ ▲ ▲ ▲ ▲ ▲ ▼ ▲		+ + + + + + + + + + + + + +		▲ ▲ (▲) (▲)	Sag: ▲ Sag: ▲	Condyles ▼ Condyles ▼	(T-L Kyphos) T-L Kyphos (T-L Kyphos) T-L Kyphos T-L Kyphos	+ + + + + + +	Hooked Hooked + Hooked Early Hooked Later Irregular Hooked	(+) (+) + (+) ++ + ++		
IX	METABOLIC BONE DISEASE HYPOPHOSPHATASIA HYPERPHOSPHATASIA HYPOPHOSPHATAEMIC RICKETS PSEUDO-HYPOPARATHYROID:	▼ ▲ ▲ ▲ ▲ ▲ ▲ ▲		(+) (+) (+) (+) + (+) (+)		▲ ▲ (▲) (▲)	▲ Sag: ▲							
X	DECREASED BONE DENSITY OSTEOGEN: IMPERFECTA IDIOPATH: JUV: OSTEOPOROSIS OSTEOLYSIS (Hajdu Cheney) (CLEIDOCRANIAL DYS:)	▼ ▲ ▲ ▲ ▲ ▲ ▲ ▲		(+) (+) (+) (+) + (+) (+)		▲ (Pat) ▼ ▲ (Pat) ▼	W ▼ W ▼	▼ W ▼	(Teeth)	(+)		Biconcave (+) (+)		
XI	SCLEROSING BONE DYSPLASIAS OSTEOPETROSIS - Severe - Mild PYCNODYSOSTOSIS CRANIO-METAPHYSEAL PYLE'S METAPHYSEAL DYS: FRONTO-METAPHYSEAL MELNICK NEEDLES ENGELMANN CRANIO-DIAPHYSEAL OSTEOPATHIA STRIATA	▼ ▲ (▲) ▼ ▲ ▼ ▲ ▲ ▲ ▲ ▲ ▲ ▲ ▲ ▲ ▼ ▲ (▲)		+ + + + + + (+) (+) (+) (+) (+) (+) (+) (+) (+) (+)		▲ ▲ (▲) ▲ ▲ ▲ ▲ ▲ ▲ ▲ ▲ ▲ ▲ ▲ ▲ ▲ ▲ (▲)			W ▼ W ▼ W ▼ W ▼ W ▼ W ▼ W ▼ W ▼ W ▼ W ▼		Infection Infection ▲ Teeth			
XII	TUMOUR-LIKE DISORDERS Diaphyseal aclasia, Ollier, Maffucci, dysplasia epiphysealis hemimelica, fibro dysplasia (myositis) ossificans progressiva, polyostotic fibrous dysplasia, melorheostosis: nearly always					frontal ▲								

† - "Lethal dwarfs" - see separate sheet: "increased limb length" - omitted  
▲▼ - increase / decrease over normal  
\* - wherever skull base is involved there may be cranial nerve compression

# SKELETAL DYSPLASIAS

PRINCIPAL RADIOLOGICAL FEATURES														PRINCIPAL ASSOCIATED ANOMALIES OR COMPLICATIONS	FURTHER INVEST: Radiology Haematology Biochemistry Genetic
Other	THORAX				PELVIS				LIMBS				OSSIFICATION		
	Cage	Ribs	Clavicle	ILIUM	ACETAB: Under-Developed ▼ Protrusio: ▲	Epiphysis	Metaphysis	Diaphysis	Coccyra	Malalignment	WRIST HANDS	Advanced ▲ Delayed ▼			
					(▼) (▲)	+	(+) Insary: dumb-bell		(+)	+	(+)	▼		Cleft palate: Myopia: Hearing ▼	(G)
Coronal Clefts						(+)	(Cupped)				(+)	Punctate Calcif: ▼ (Hip)		Cataract; heart; skin lesions	B.
Post. Vert. lump	Carinatum			▼		+ (less early)			++		(MC ▼)	▲		Cleft palate; Myopia	R. (Cl.2) (G)
		▲ ant						+		+					B. (? rickets)
		Infant ▼				▲	++	(+)	+	+		▼		Blood ▼, Hair ▼	B.H.
				▲			+	(+)	+					Hearing ▼, Fractures	B.
							+	(+)						Cyclical neutropenia	B.H.
Pedicles ▼	(▼)	(▼)		▼	Spur		+	(+)	(+)	Tib / Fib Tib / Fib	+			Spinal stenosis	G.
Pedicles ▼							(+)			+	Dist. ulna (Madelung)				G.
Pedicles ▼							+			+		▲ Hands			(G)
Pedicles ▼		Post cupping		▼	▼	++	++		(+)	Contracture	▼ 1st MC	▼ (Hips)		(Gross joint laxity)	(G)
Pedicles (▼)				(▲)		+	+		(+)	Contracture		▲		Cleft palate	R. (Cl.2) G.
Coronal Clefts				▼	▼	(+)	Dumb-bell	▼		+		▼		Cleft palate; myopia; hearing (Mental retard.)	(G)
		Carinatum		Lace like		(+)	▲			(+)		▲		Heart; hair ▼	(G)
		▼ improves late ▼		▼	Spike	(+)	+			(+)	Fusion coxarth Post-ex. poly.	(▲) Infant		(Nephritis)	(G)
		▼ improves late ▼		▼	Spike	(+)	+			(+)		(▲) Infant			(G)
		All May: } Med: ▲ Med: (▲) Ant: ▲ Post: ▼ Carinatum }		▲ ▲ (▲) (▲) Med: ▲ Med: ▲ Med: (▲)	▼ ▼ (▼) (▼) (▼) (▼) (▼) (▼)	+	+	▲	(c. valga)	Contracture	MC 2-5 pointed bases	Modelling ▼		Mental retard:	B.G.
						+	+	▲	(c. valga)	Contracture	As Hurler	As Hurler		Mental retard:	B.G.
										Contracture	Contracture	Contracture		Mental retard:	B.G.
										Contracture	Contracture	Contracture		Mental retard:	B.
										Contracture	As Hurler	As Hurler		Mental retard:	B.G.
						+	+	▲	(c. valga)	Contracture	As Hurler but well modelled	▲ Hip-early ▼ Hip-late		Joint laxity; Atlanto-axial instability	R. (Cl.2) B. (G)
								+	(c. valga)	Groin gump ++	As Hurler			Periodical creaking	R.
							+	(+)		+	+	▼		Fractures	B. (G)
Pedicles ▼		▲ Dense 'rosary'			▲		Cupped		(+)	+	+	Modelling ▼		Fractures	B.
										+	+	▼ prem. fusion		Fractures	B. (G)
												MC 3, 4, 5 ▼		Hypocalcaemia	B. (G)
		(Beaded)		(Trefoil)	▲					+				Fractures (Blue Sclera) (Dysthes)	(G)
														Fractures	
Fusion delayed		▼ Thin		▼ (Pubic synph: wide)	(▲)					+	T.P. ▼	▼		Fractures (Pseudarthroses)	(G)
Sandwich		(▲) (▲) + (▲)		Arcuate bands Arcuate bands		▲					'Bone-in-Bone'			Fractures; Pancytopenia (Fractures)	(G) (B)
														Fractures	(G)
								▲ (early) ▲			T.P. ▼	Modelling ▼		Fractures	
											Modelling ▼	Modelling ▼			
		▼ Constrict. ▼ Constrict. (▲)		Grooved ▼ Grooved ▼	▲			Constrict: ▲		Wavy Wavy	Modelling ▼	T.P. ▼		Cosmetic only	(G)
											Modelling ▼			Pain; Muscle weakness	

: diagnosed over the age of 2 years; all are asymmetrical with shortening and/or malalignment confined to the affected part, with patchy sclerosis. Malignant change is rare, except in Maffucci.