The Radiology of Inherited Metabolic Bone disorders

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Outline

- Background: Skeletal dysplasias
- Lysosomal storage diseases
- Congenital Disorders of Gycosylation
- Peroxisomal disorders
- Inborn errors of cholesterol metabolism
- 2 D-glutaric aciduria
- Osteopenia

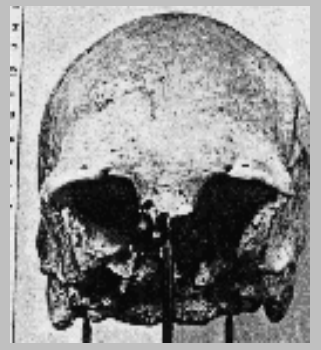
The Human skeleton

- complex organ
- 206 bones
- appendicular (126), axial (74), ossicles (6)
- functions
- 2 tissues, 3 cell types

Skeletal dysplasias

- Abnormalities in the development, growth and maintenance of the human skeleton

BONE FORMATION



ENDOCHONDRAL

- bones form from cartilage anlagen
- complex process of chondrogenesis, hypertrophy, apoptosis, bone replacement
- Major mechanism of skeletogenesis

INTRAMEMBRANOUS

- mesenchymal cells condense
- tissue vascularizes
- cells differentiate directly to osteoblasts
- Flat skull bones, clavicles form this way

CURRENT NOMENCLATURE

Am J Med Genet Jan 2007, 143A:1-18.

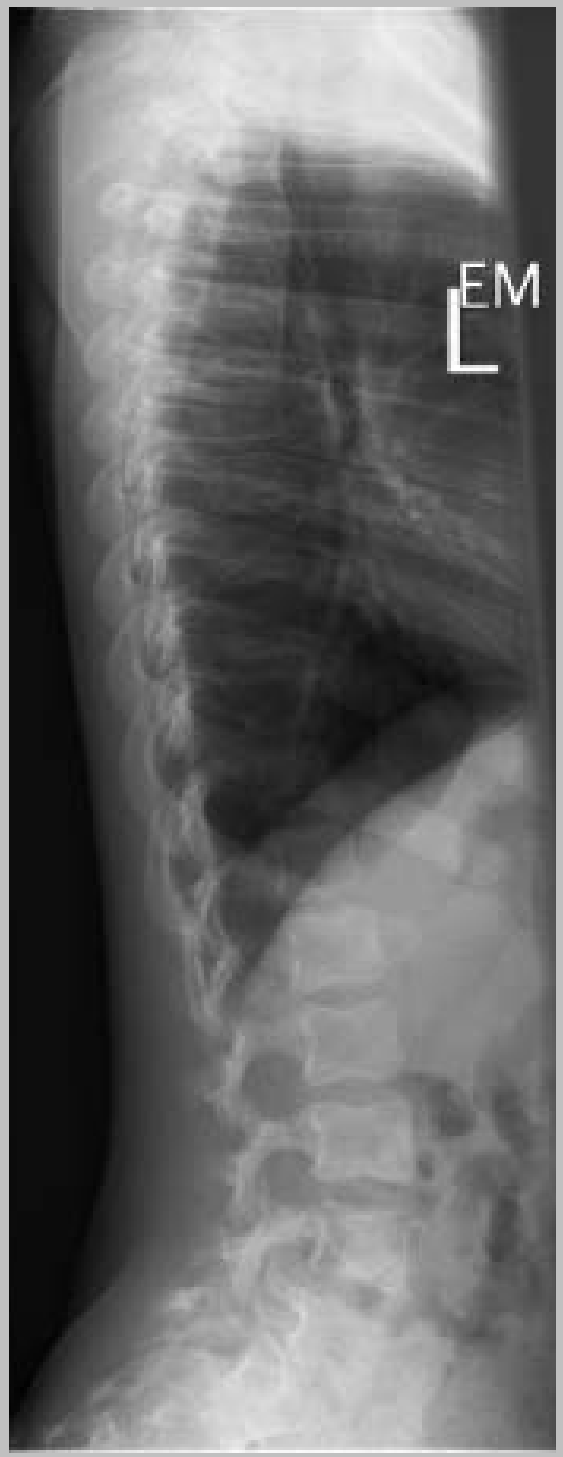
- Over 350 well defined entities in 37 “groups”
- Many groups now based on common etiology (i.e. achondroplasia group, type 2 collagenopathies, diastrophic sulfate transporter group)
- Many disorders due to inborn errors of metabolism

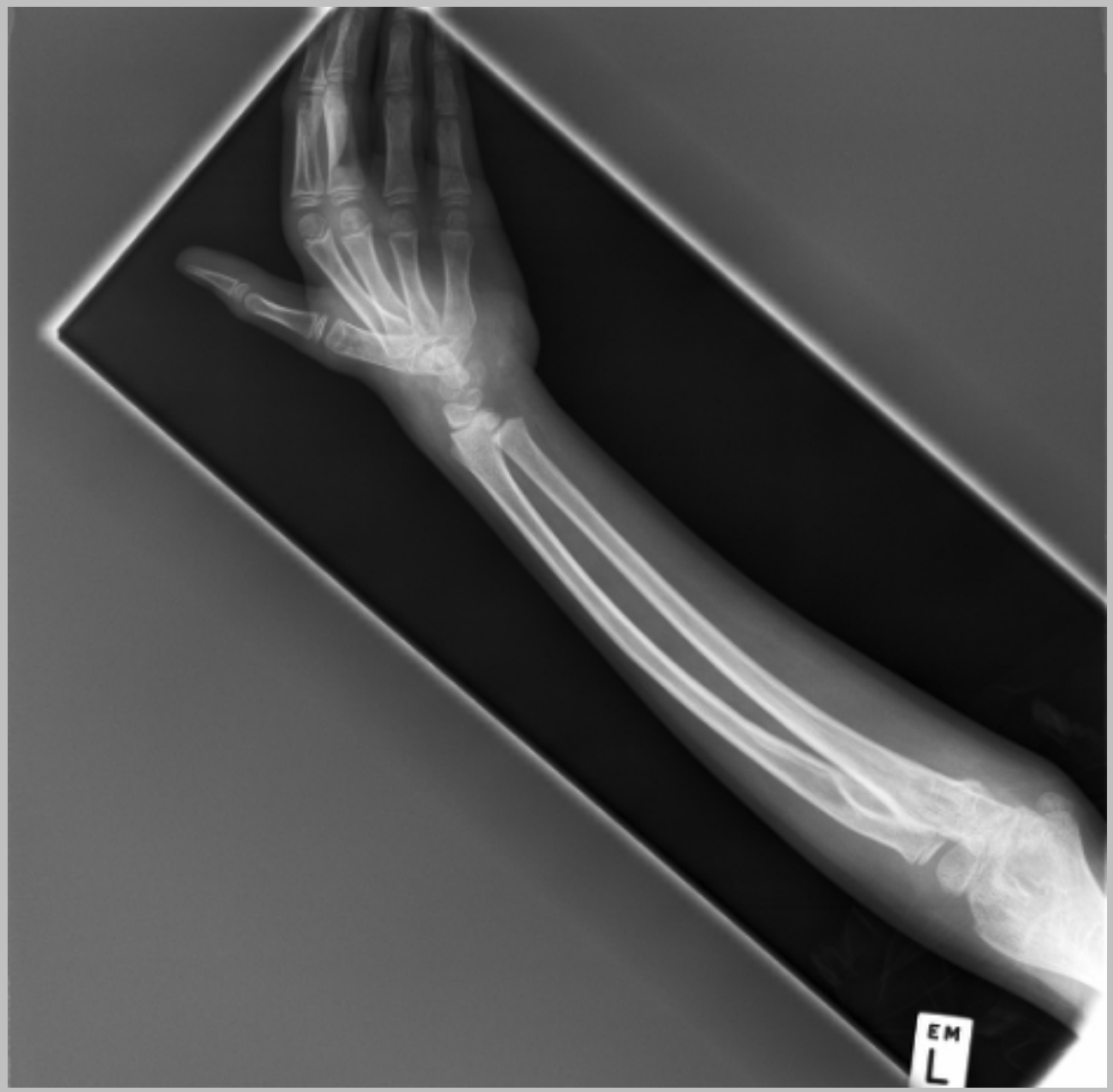
Lysosomal Storage Disease

- MPS
- MLS

- Dysostosis multiplex
- Important radiographic features

MPS I





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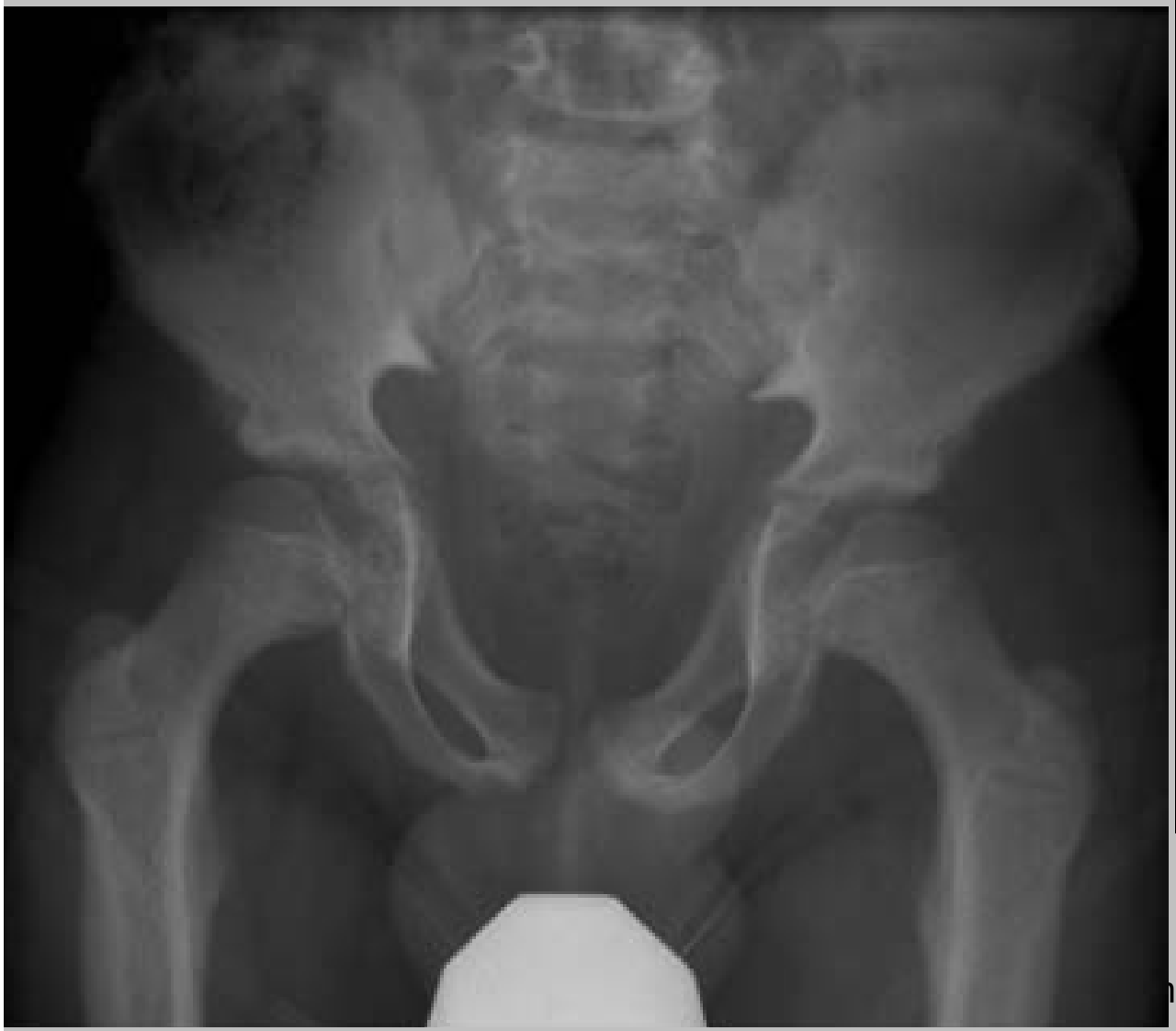

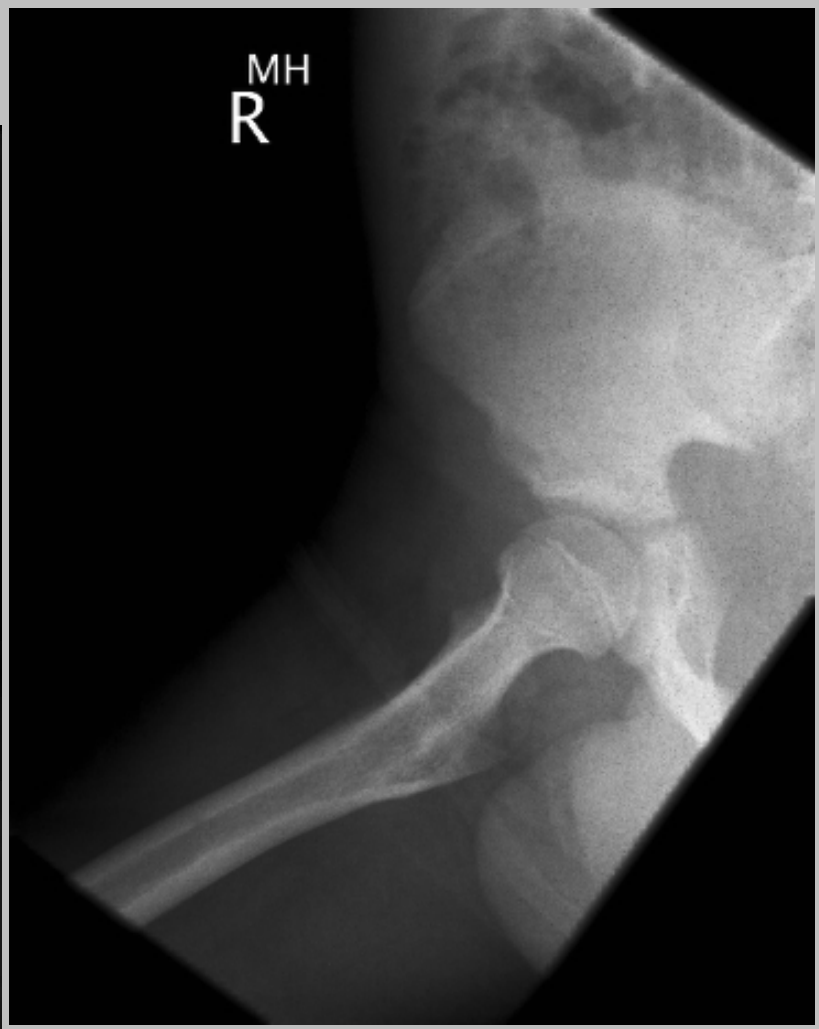

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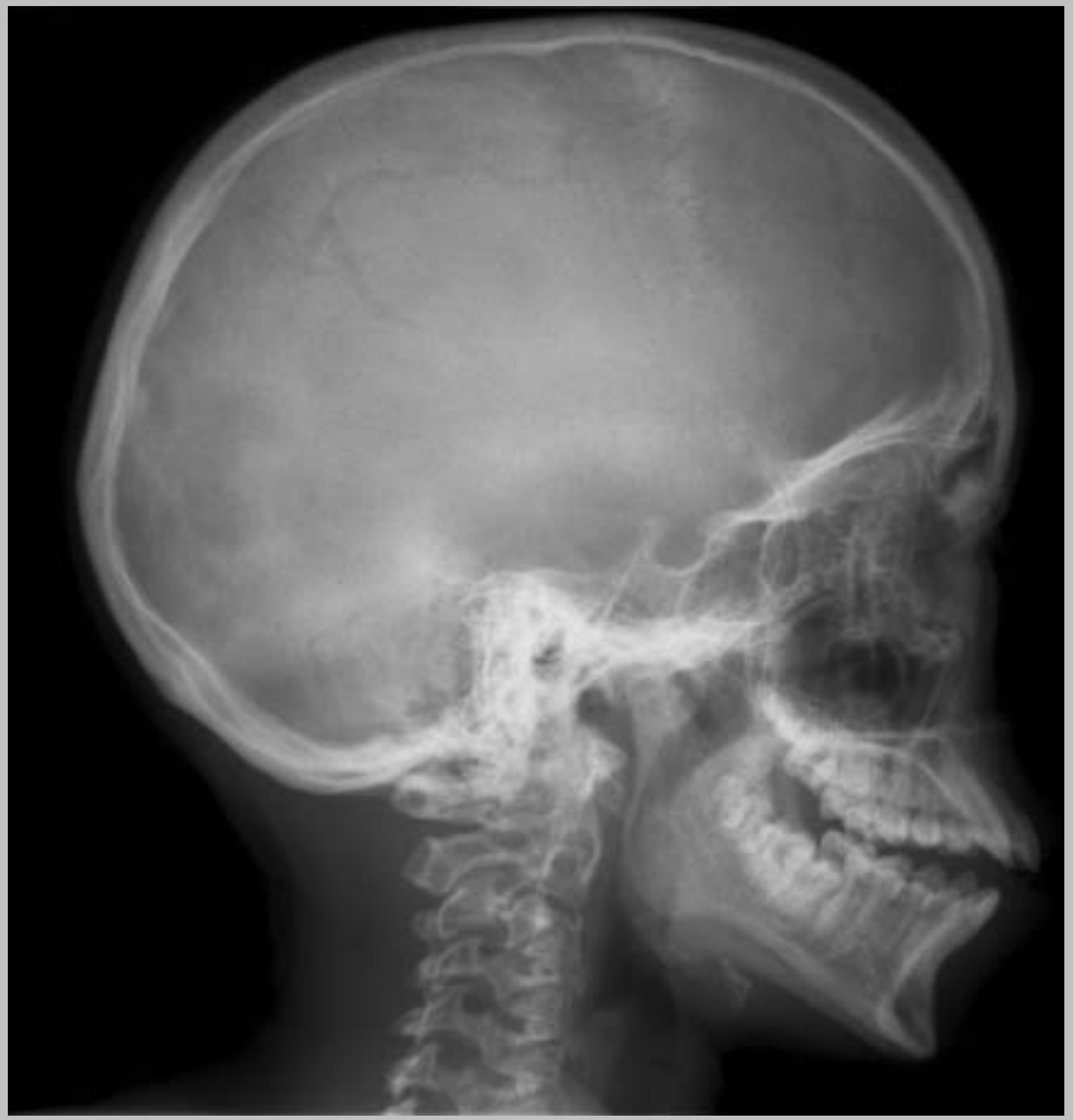

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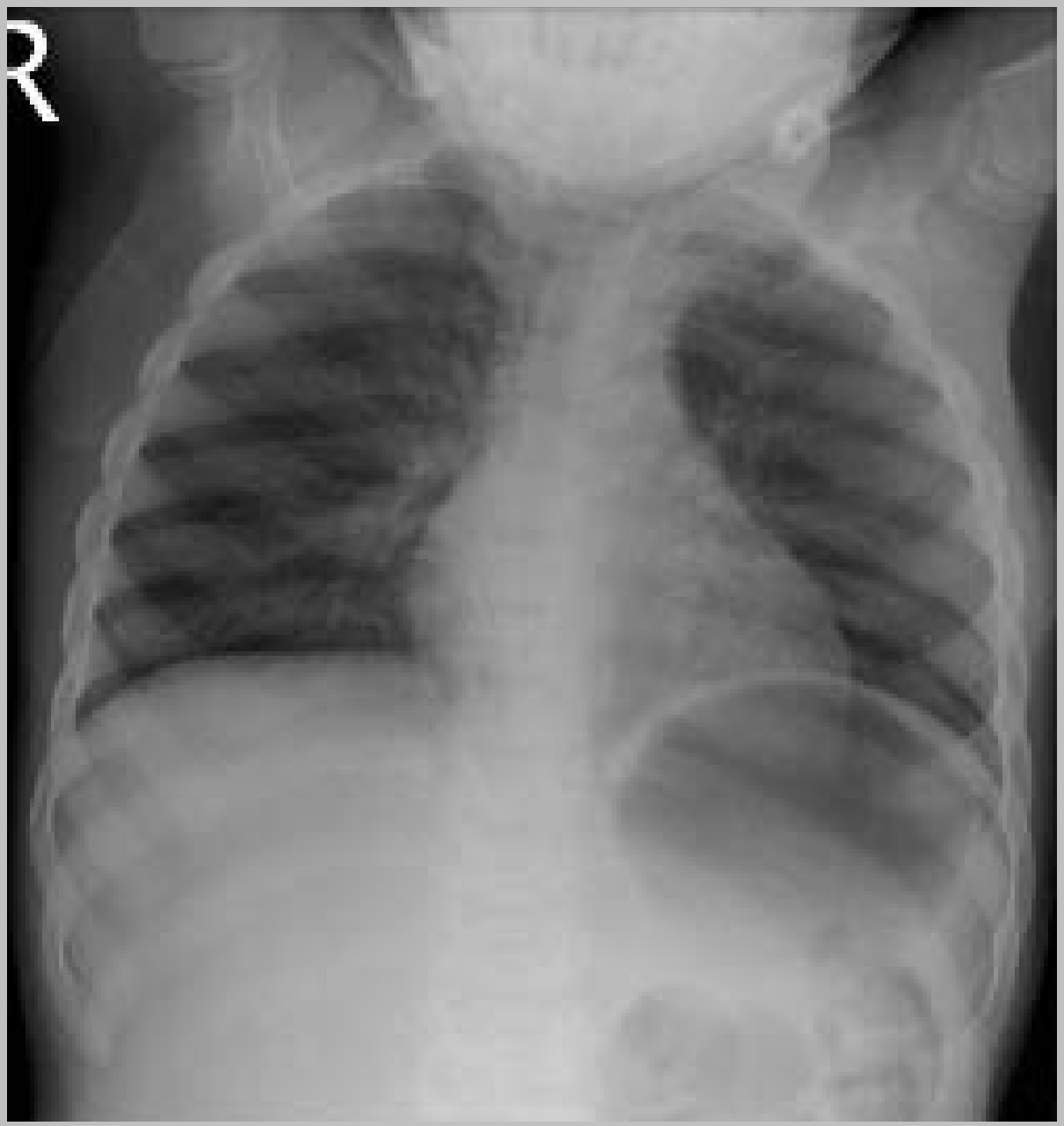

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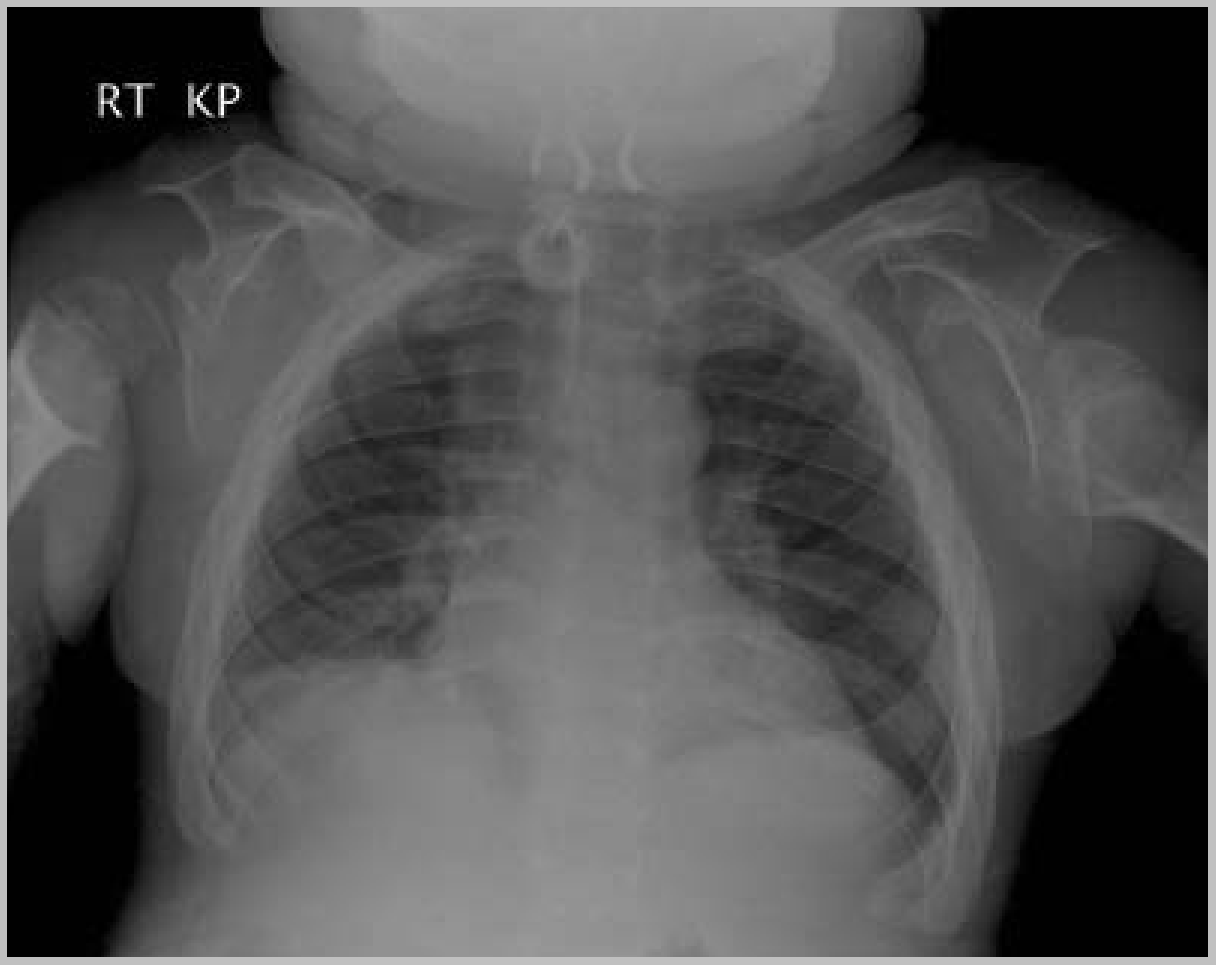
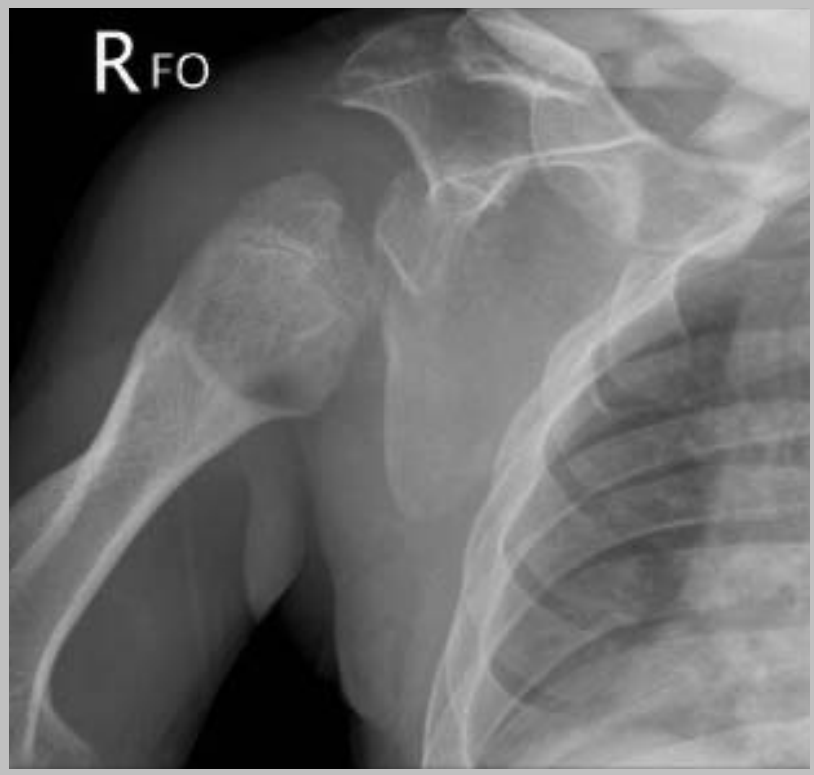
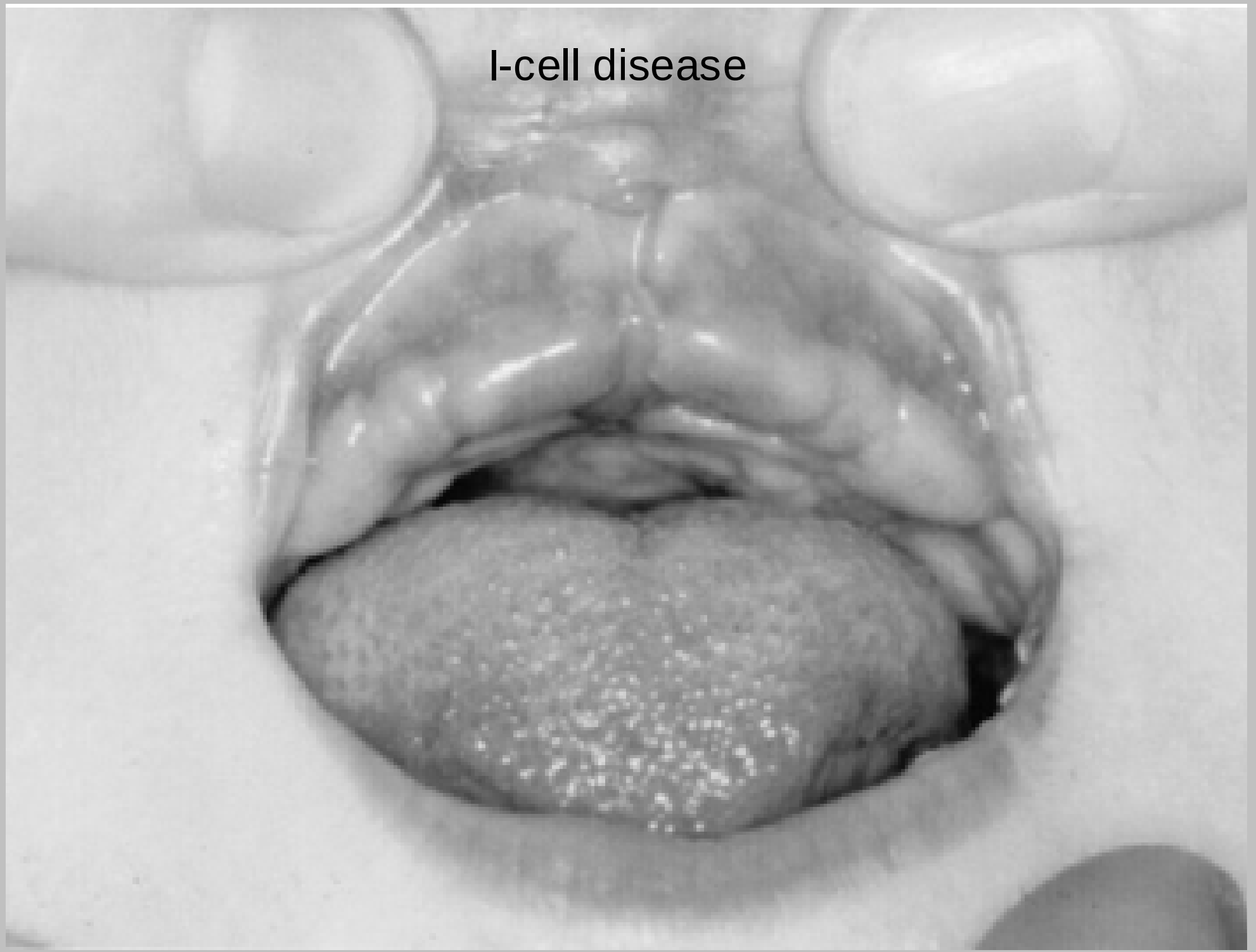
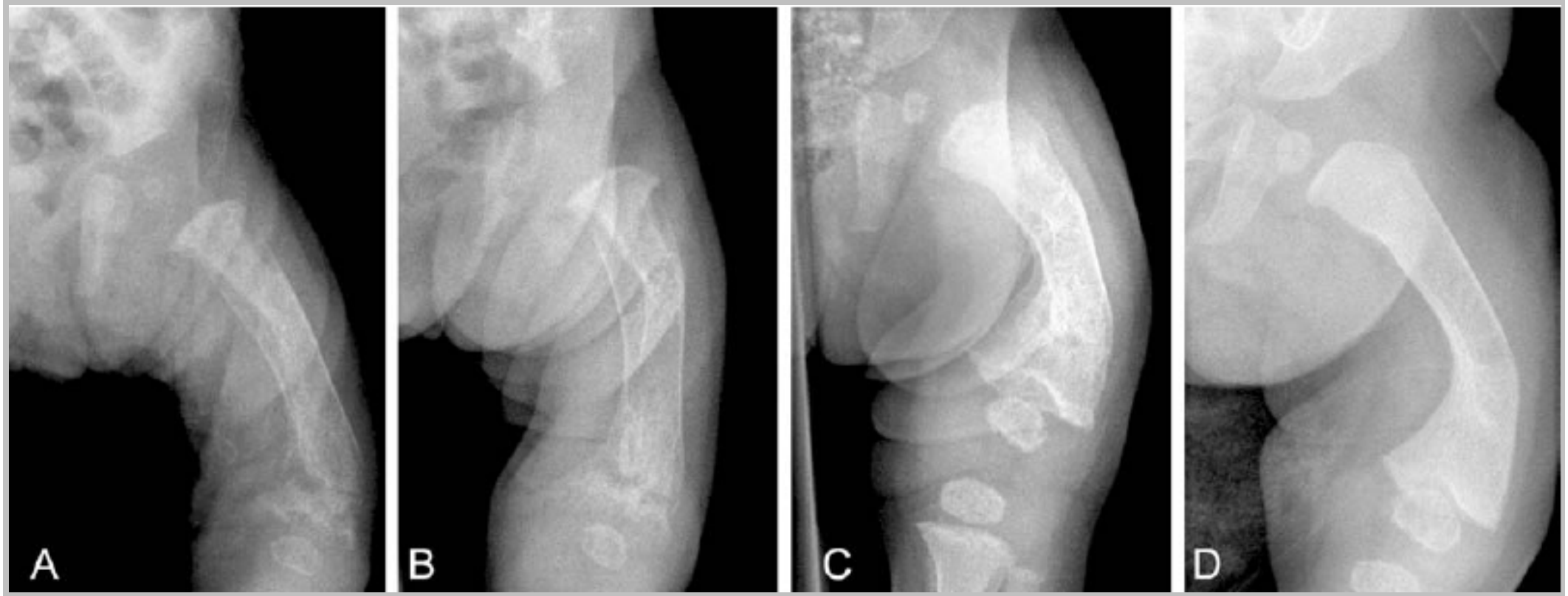




I-cell disease

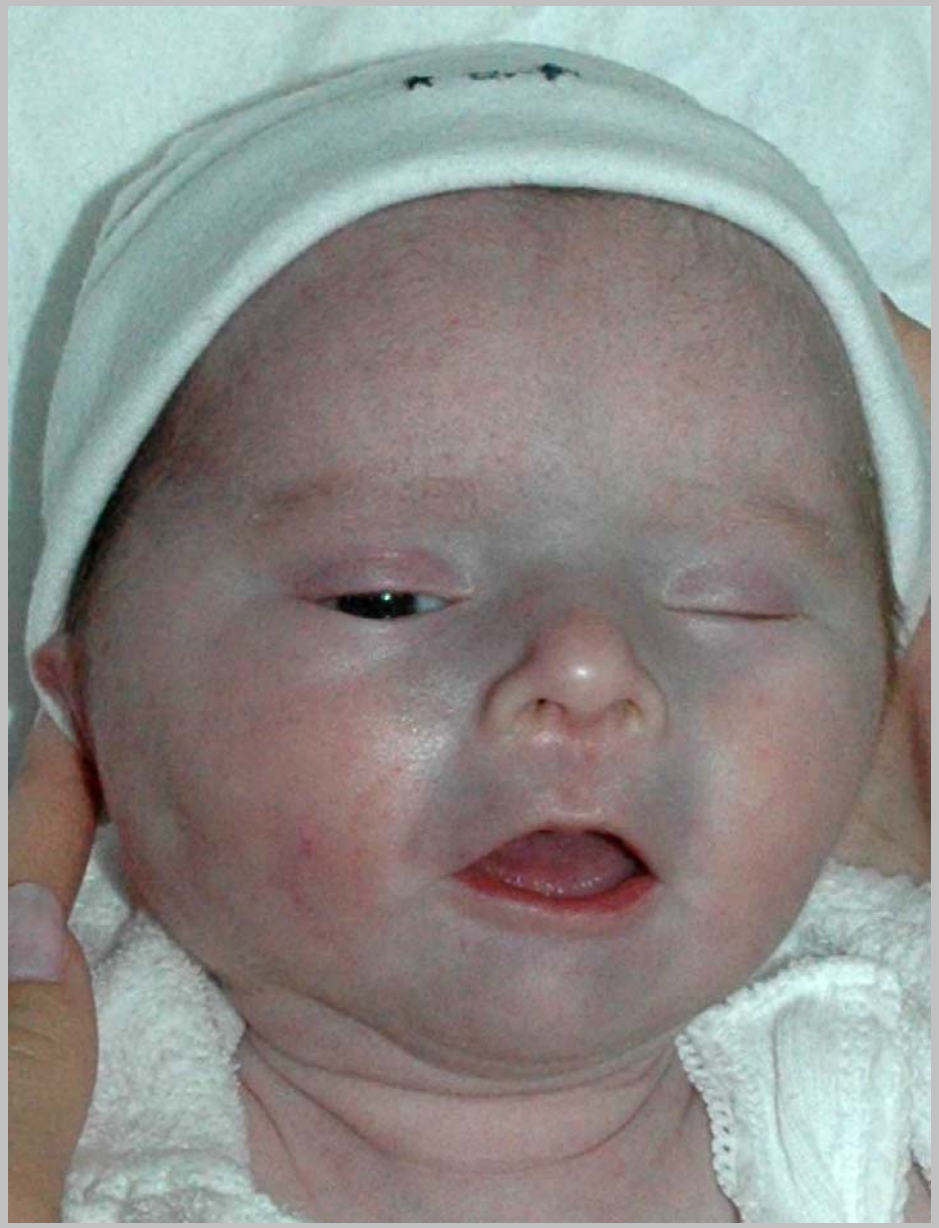




Unger et al 2005

Congenital disorders of glycosolation

CDG 1A



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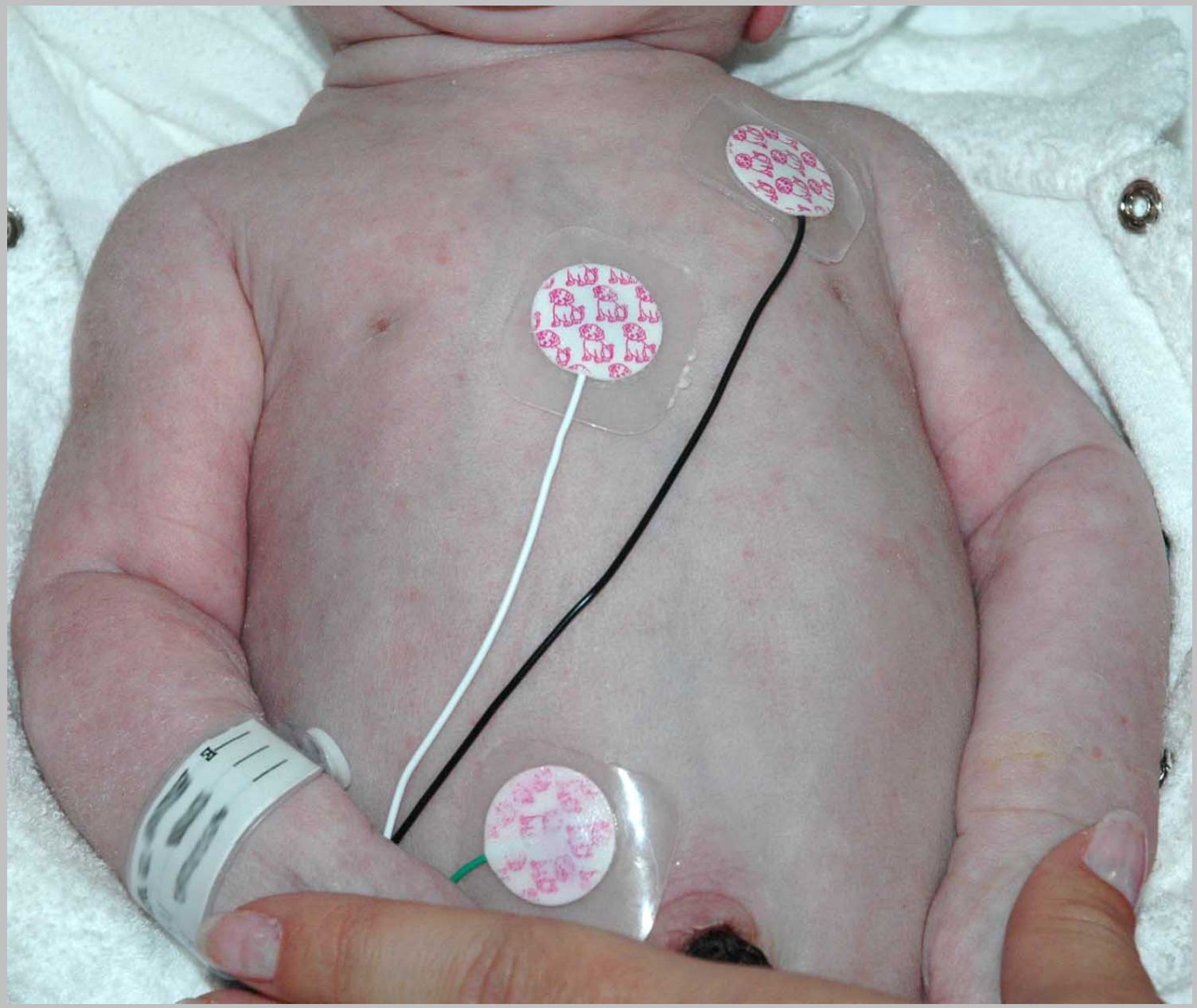

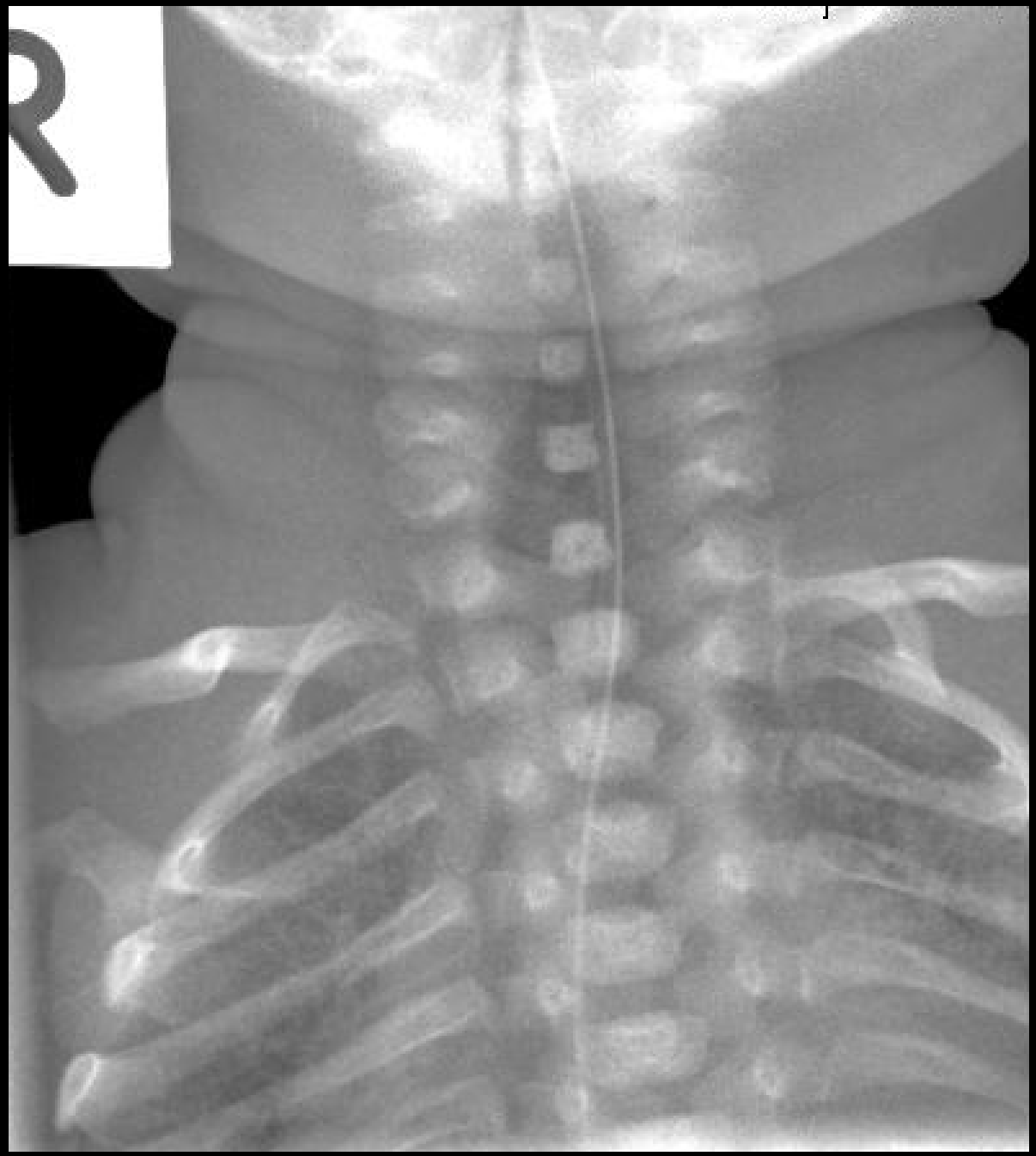
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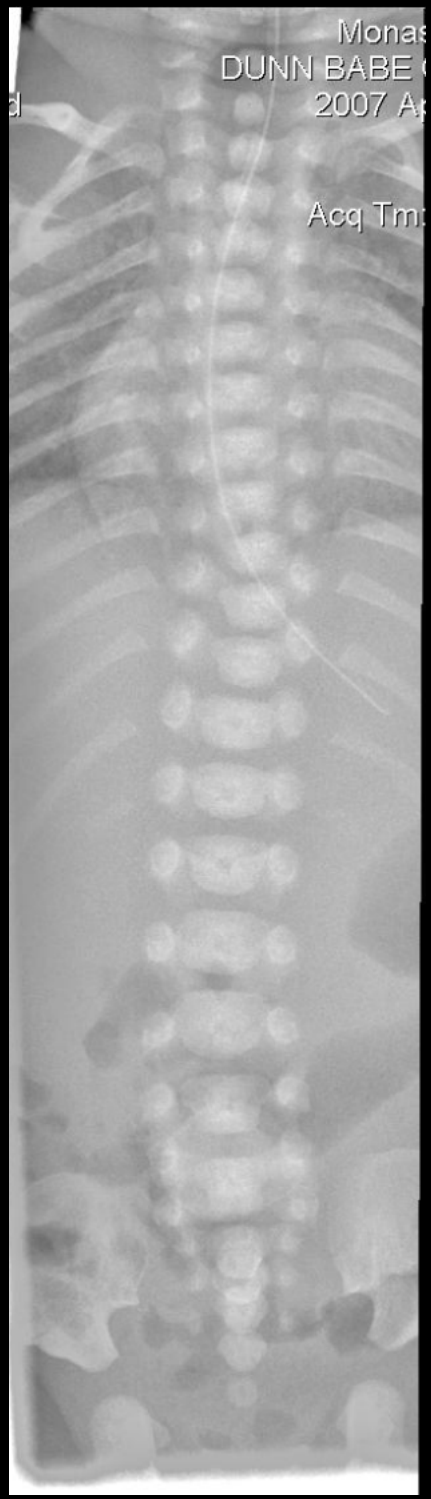
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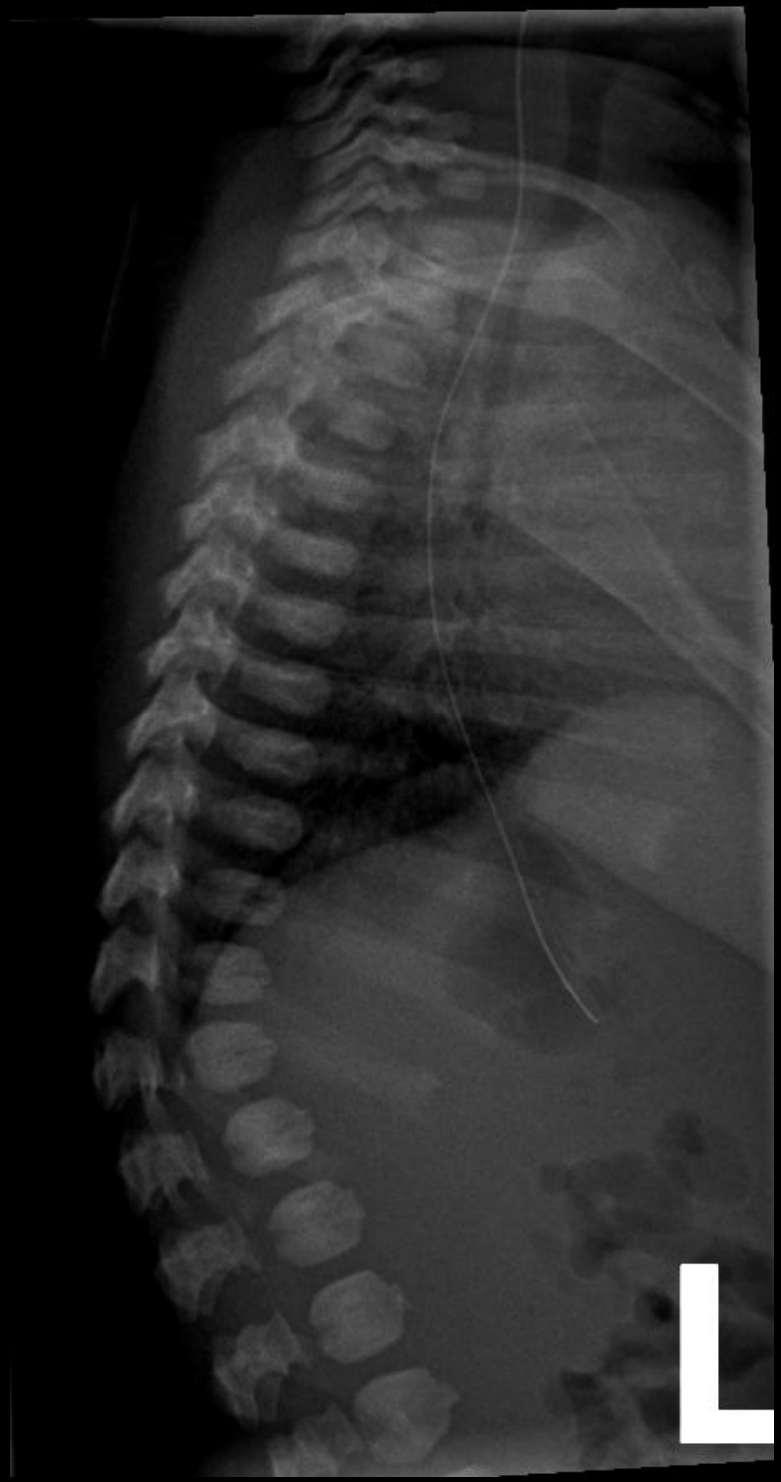
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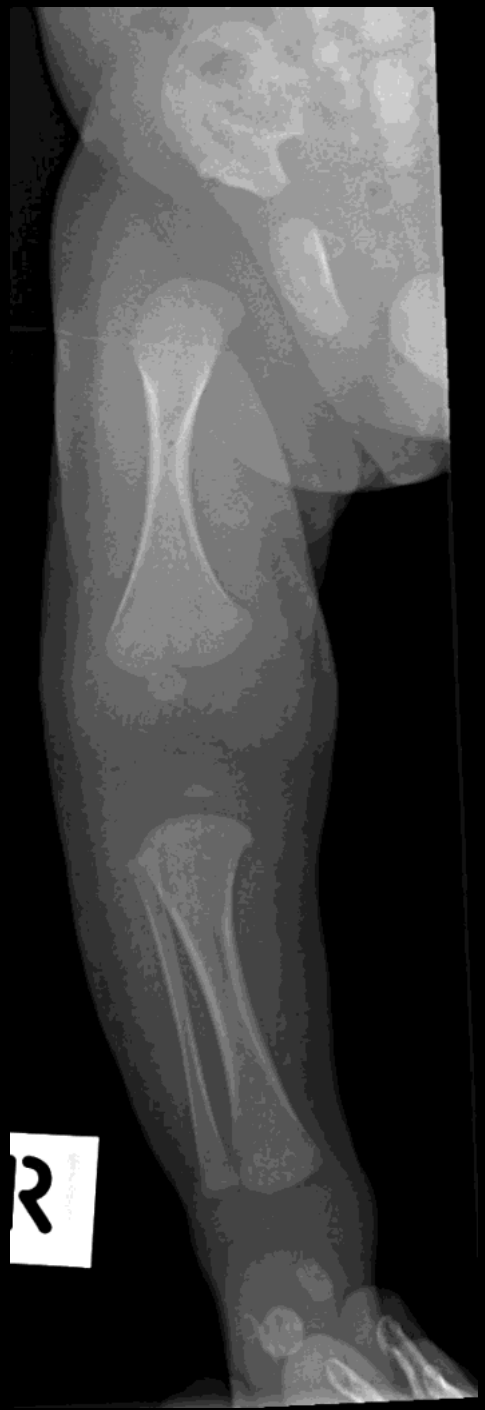
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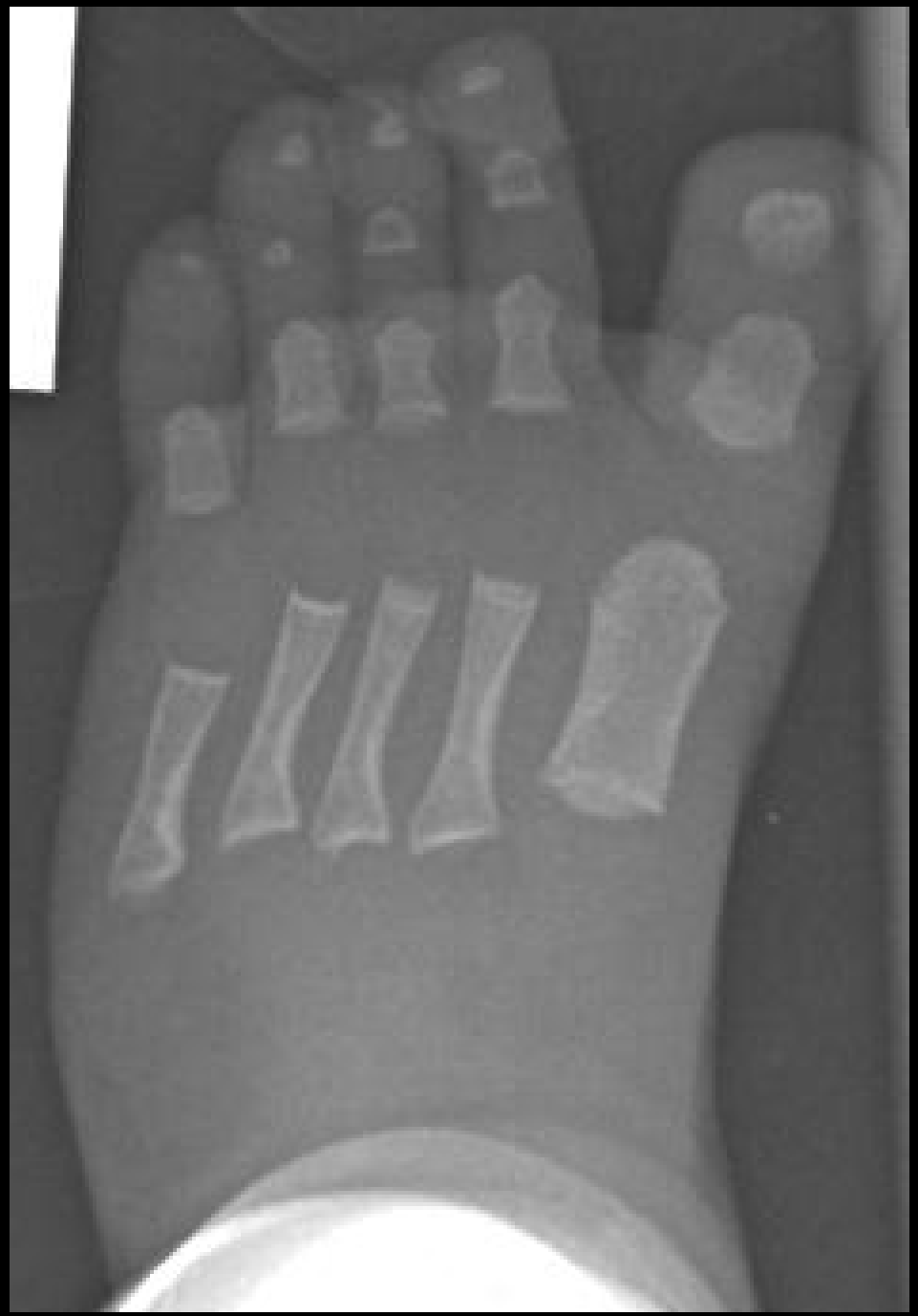

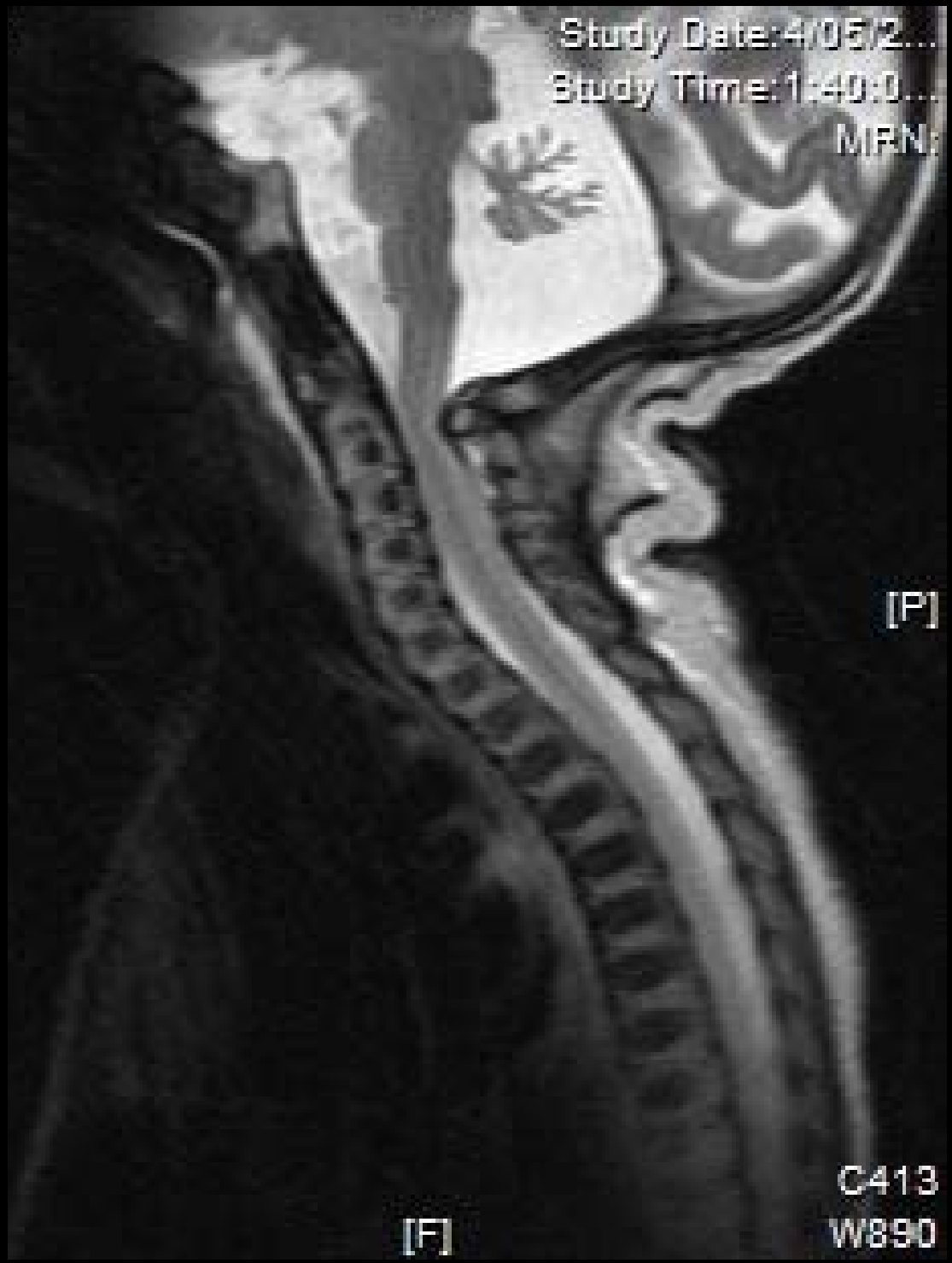
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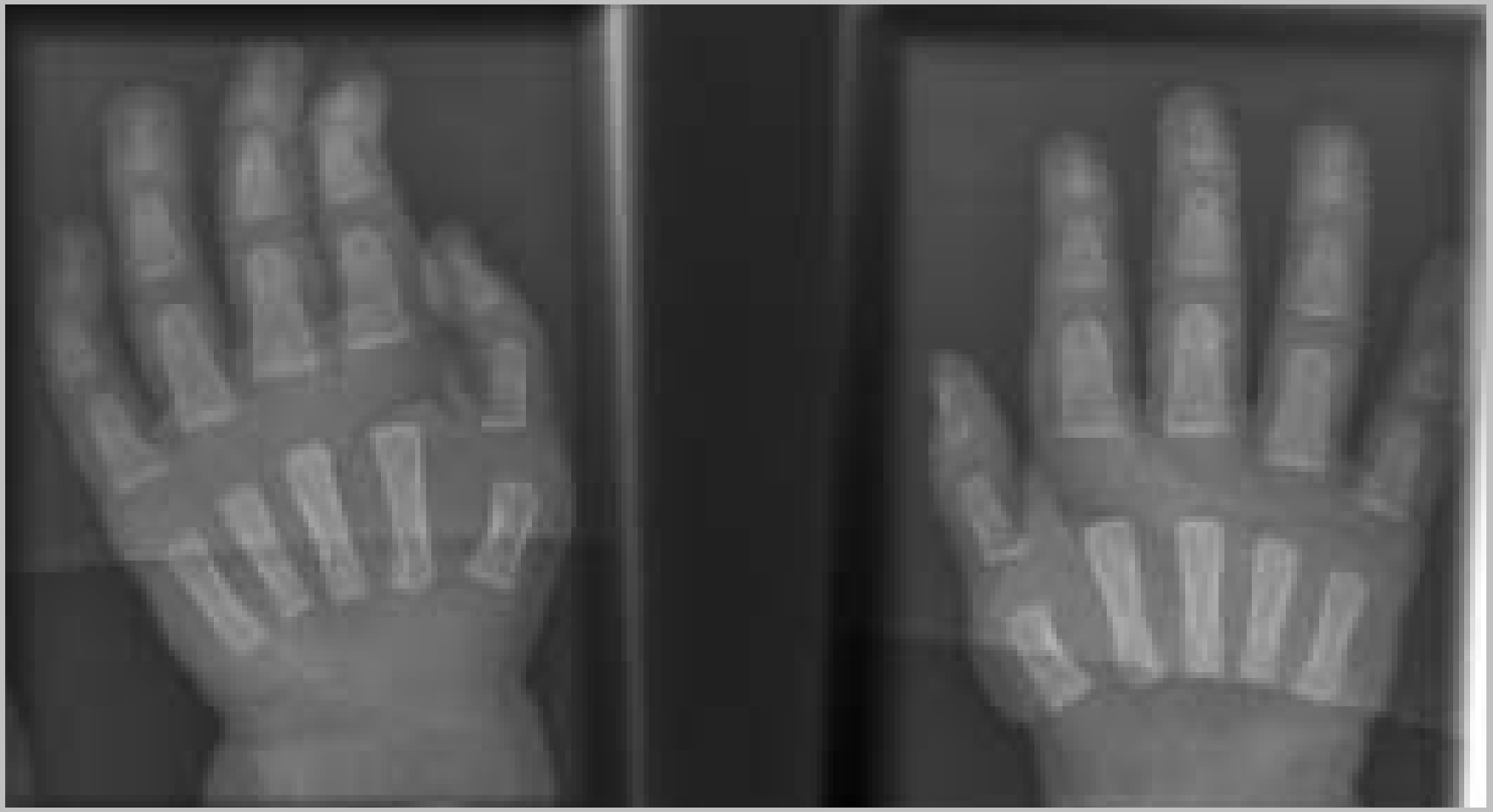

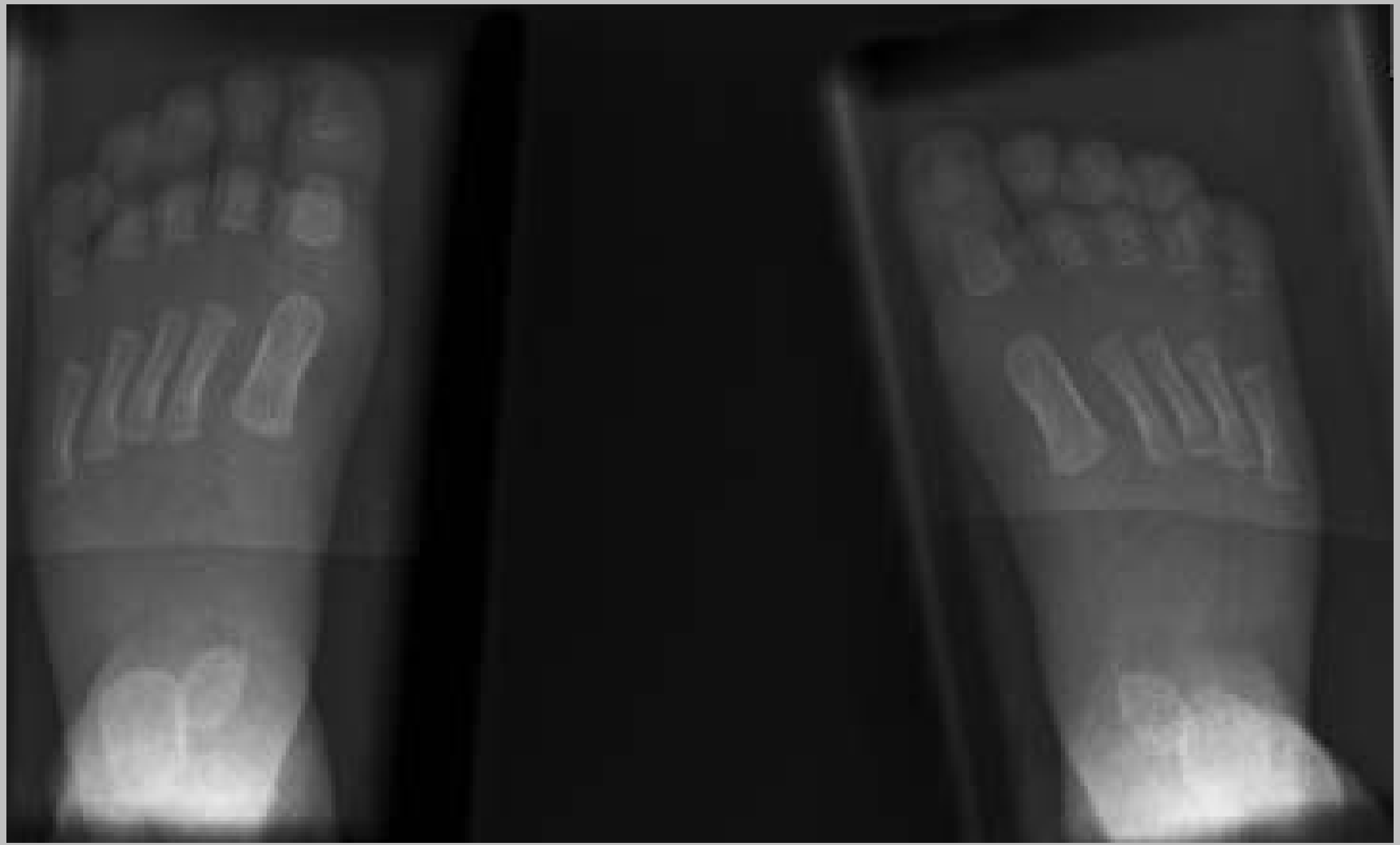

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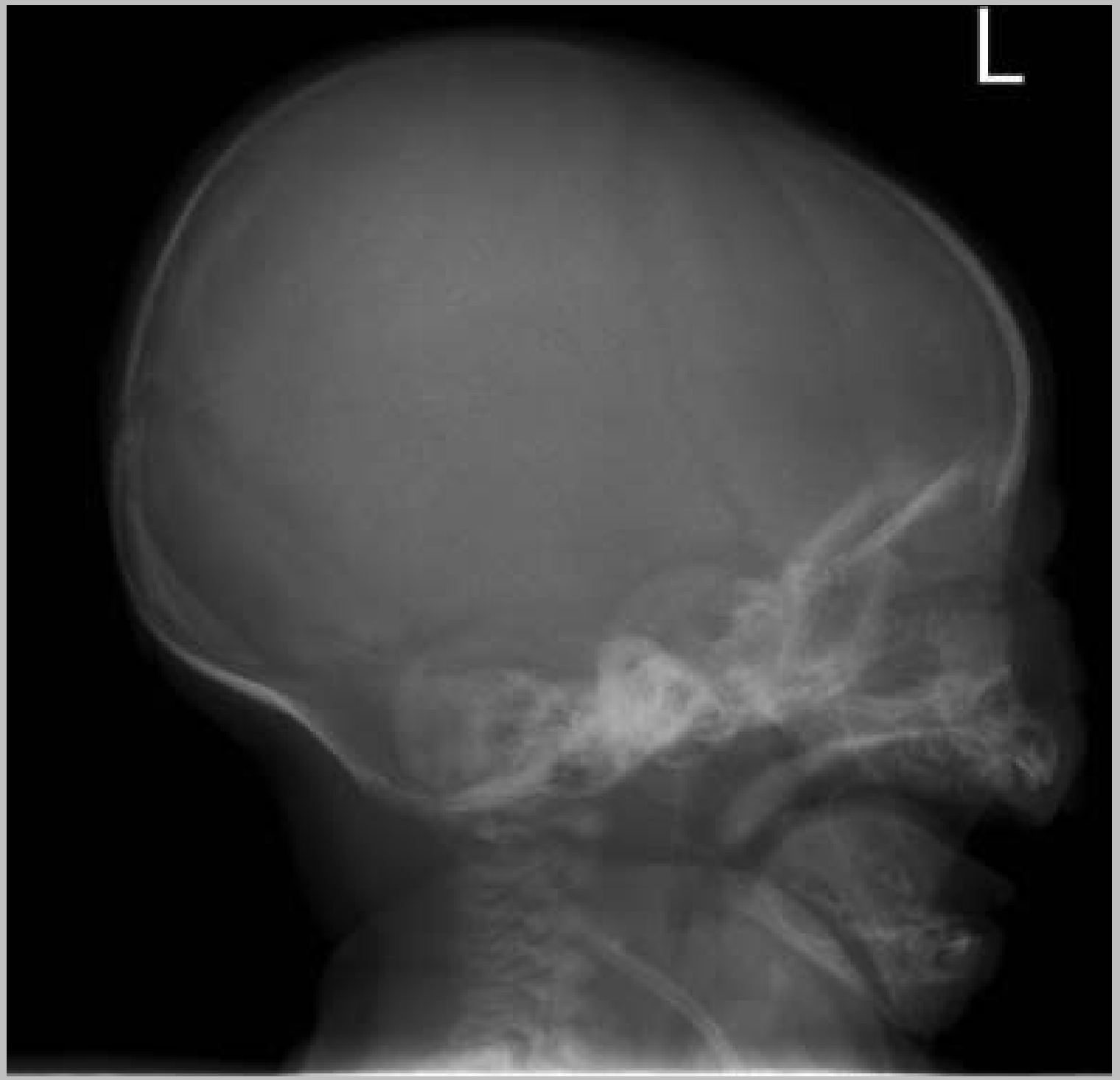

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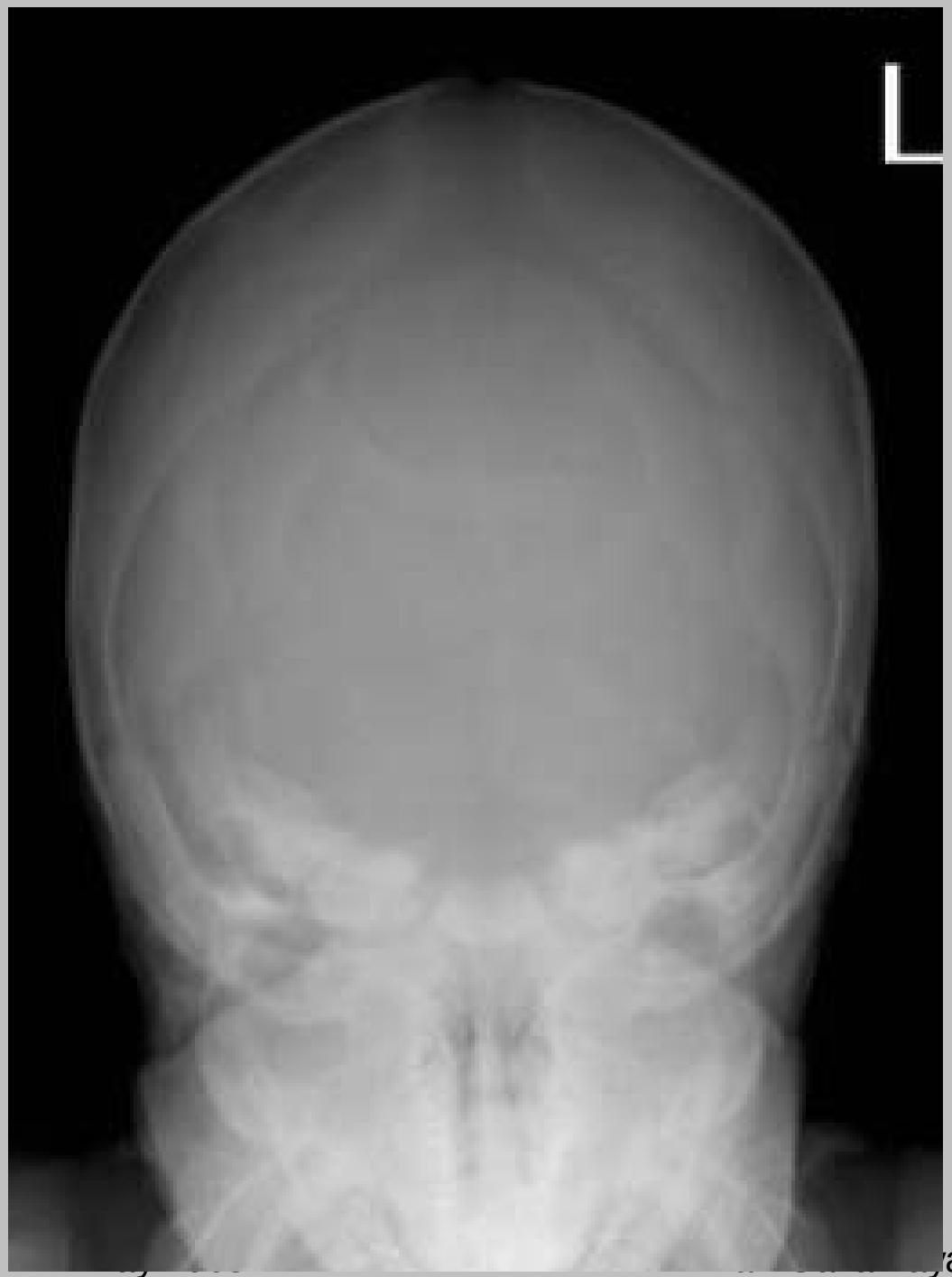
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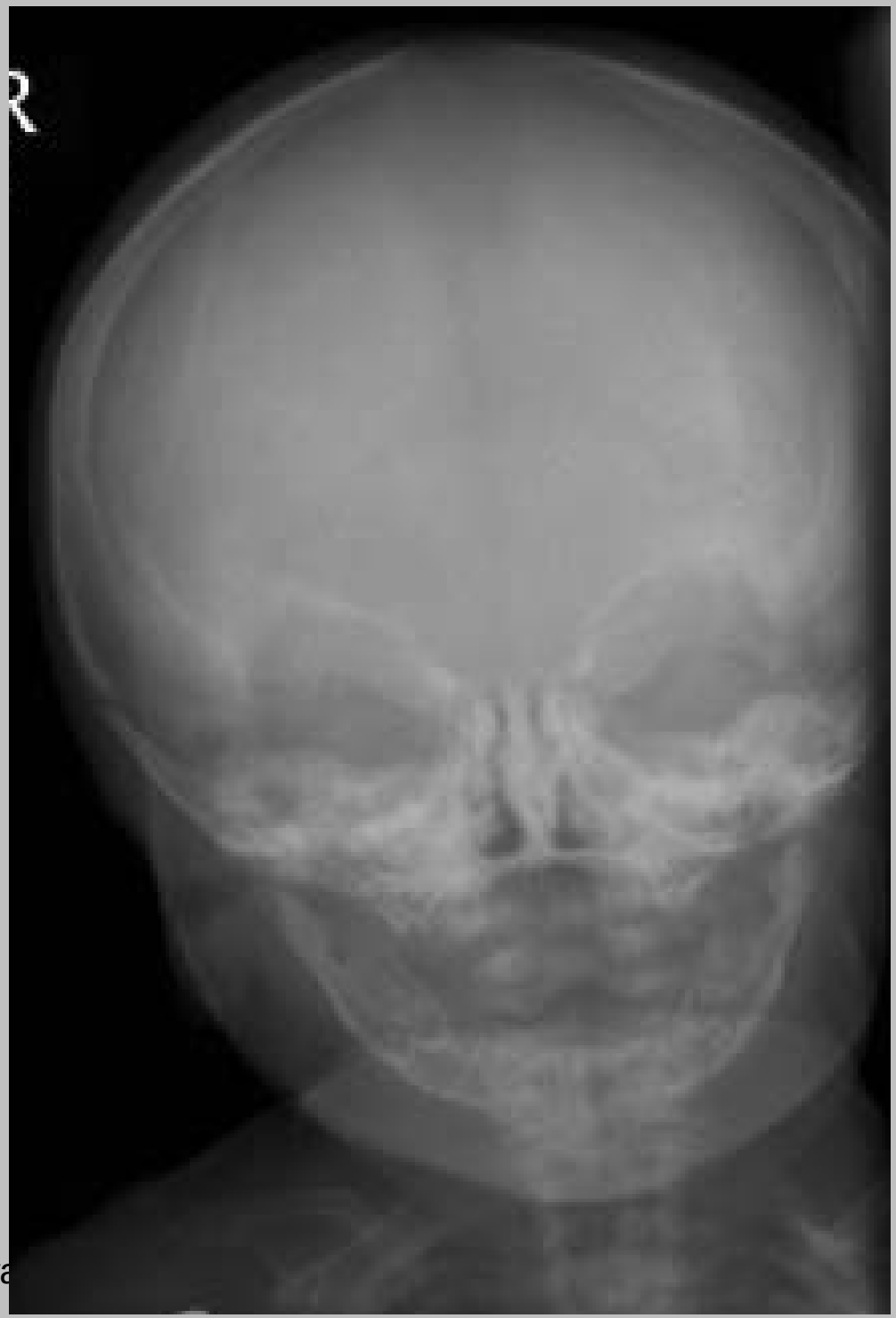
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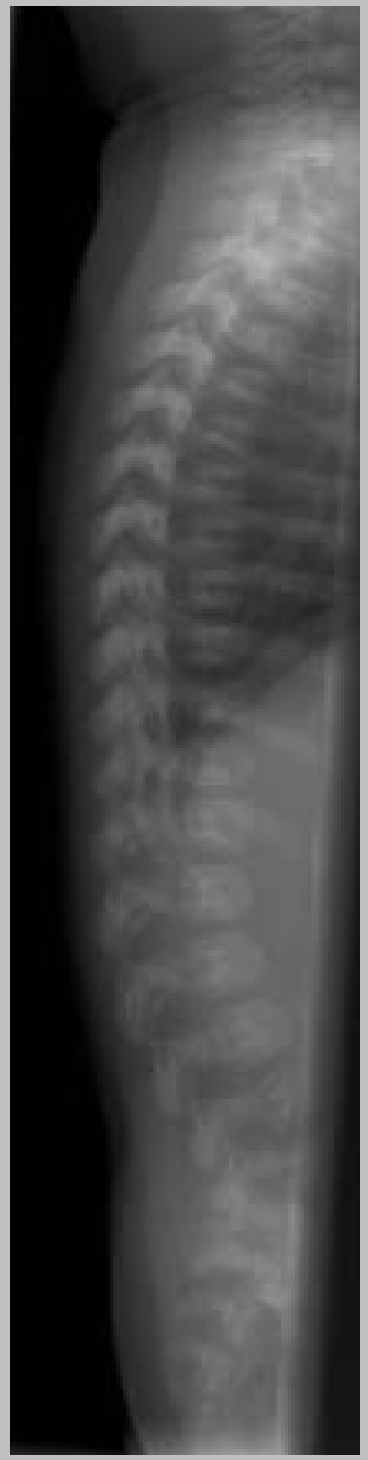

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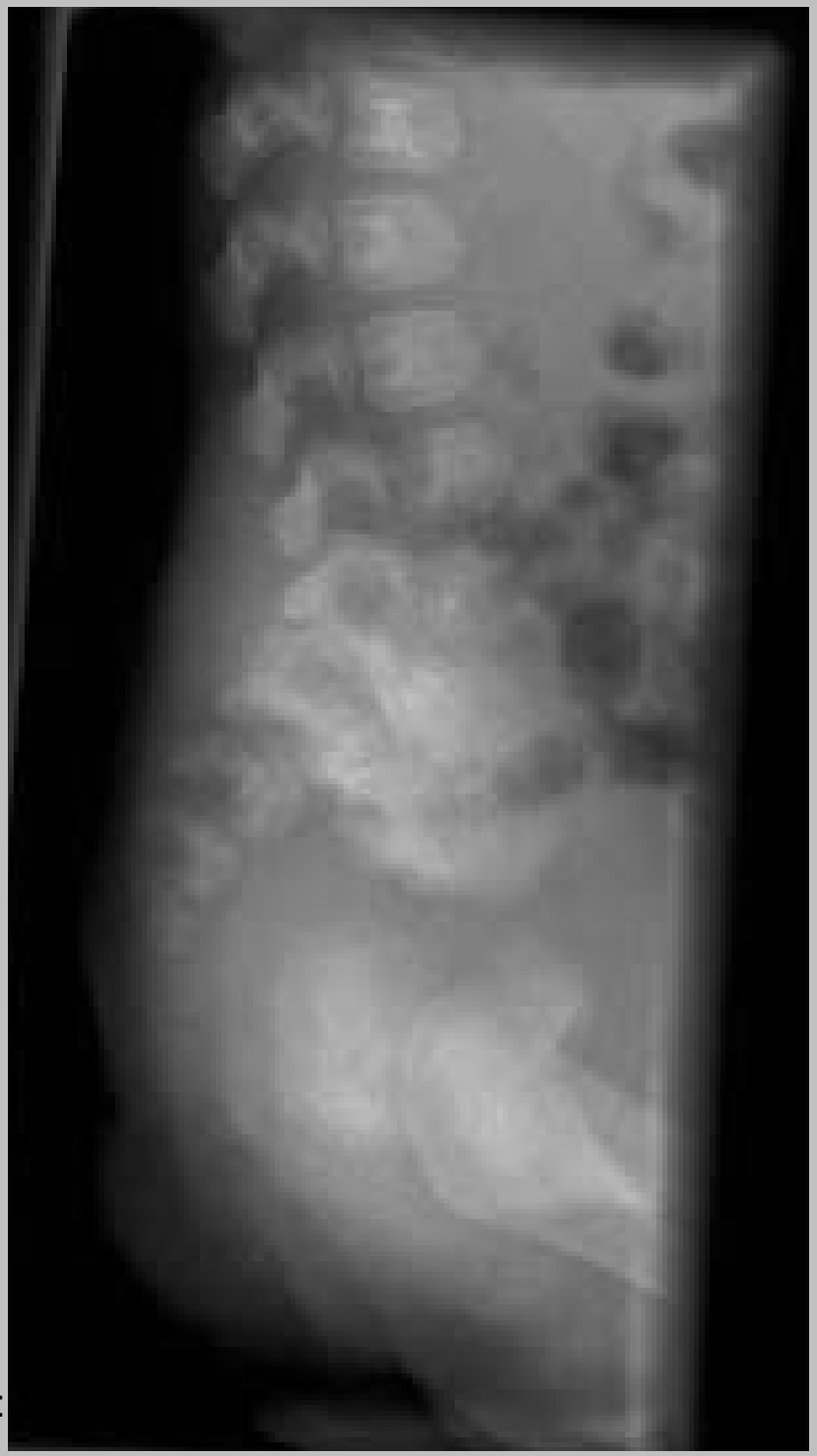

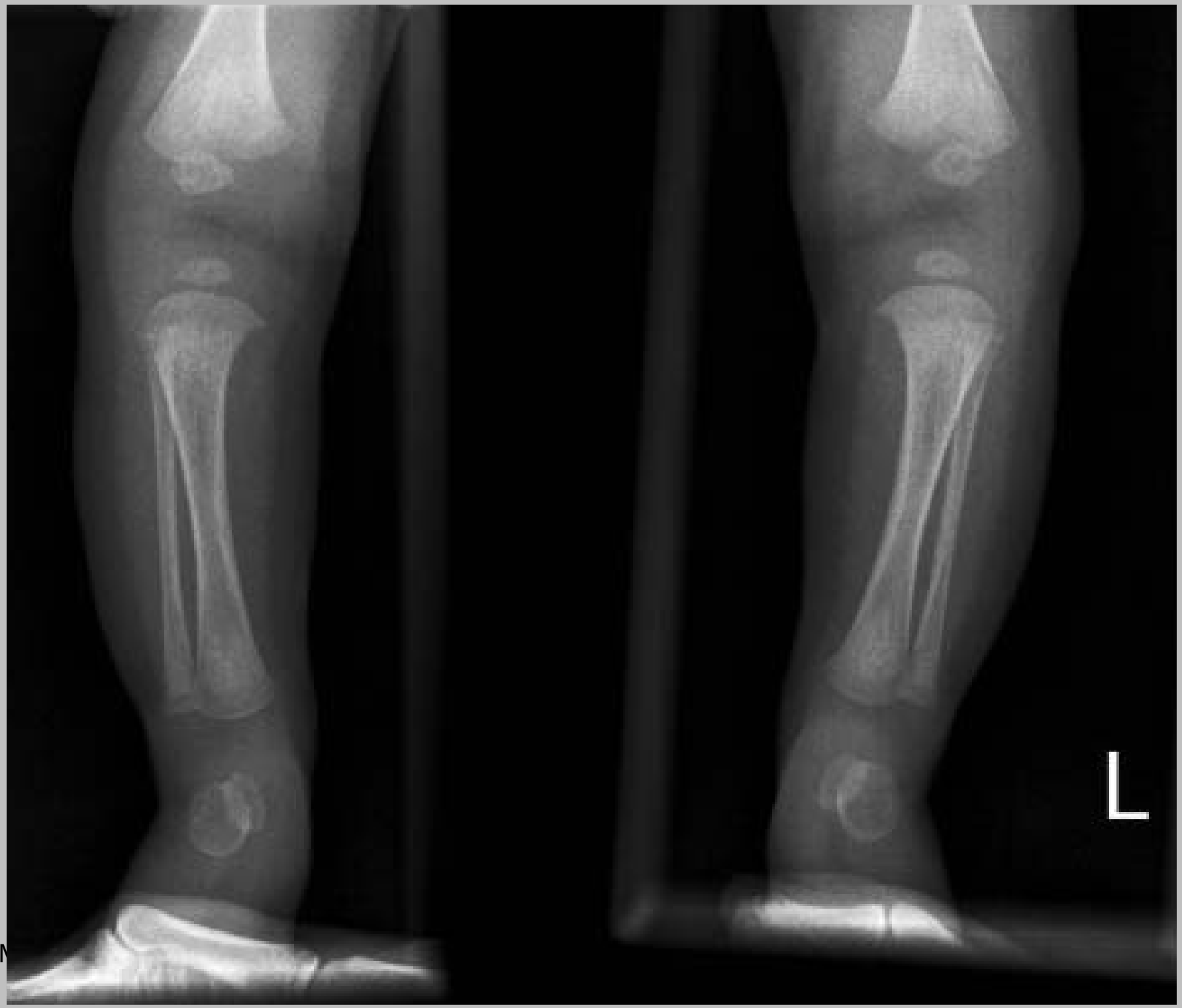
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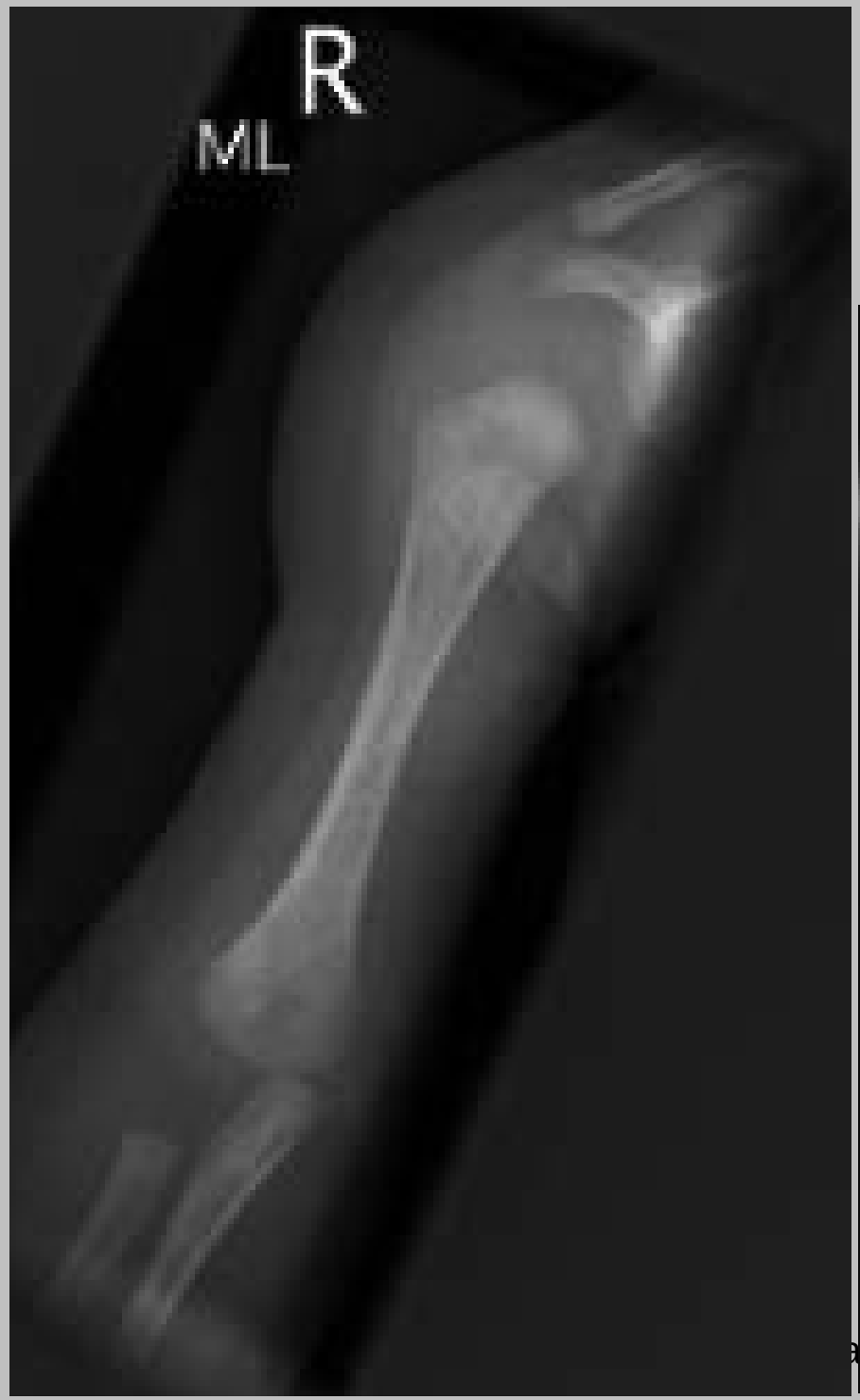
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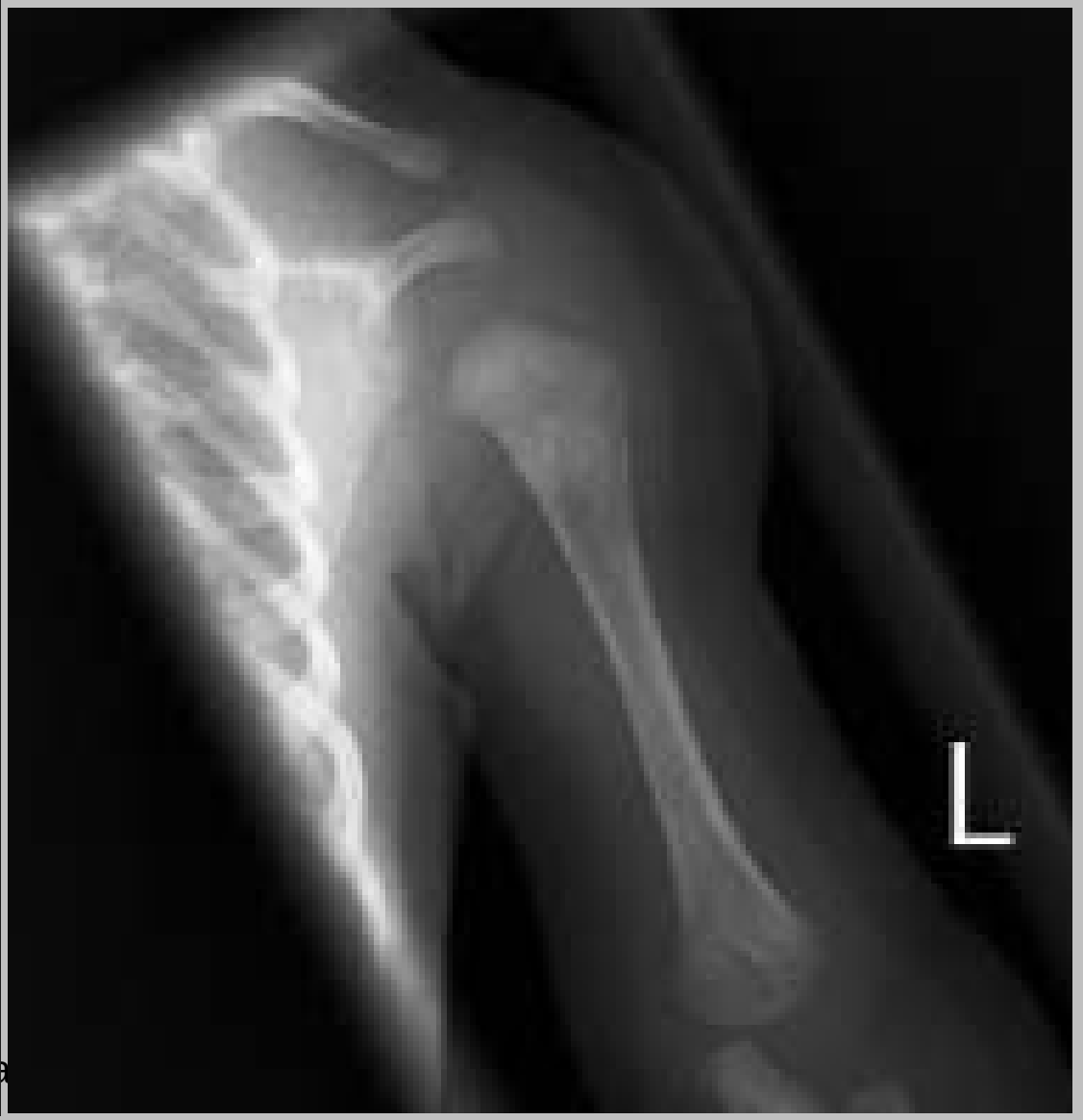
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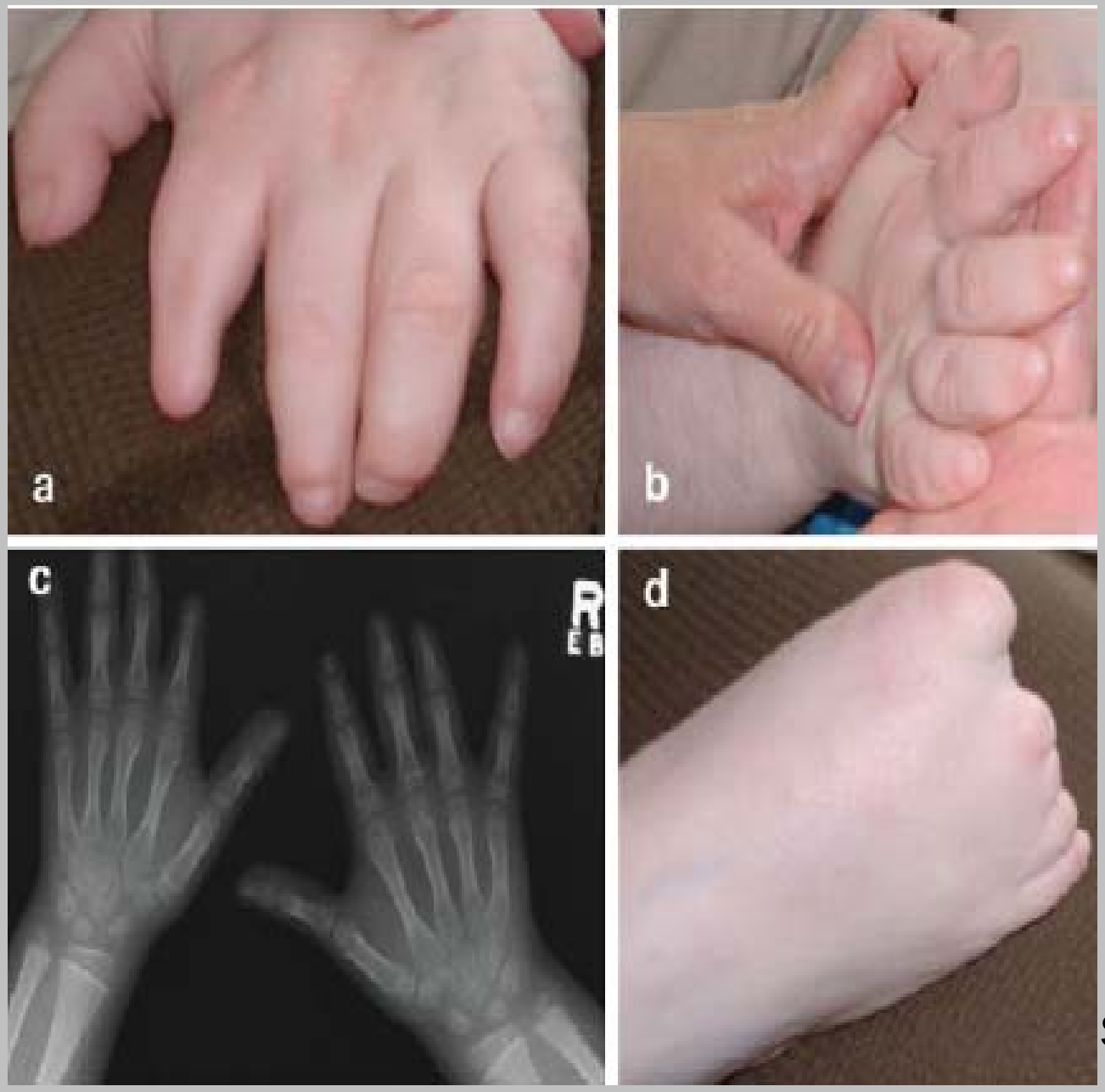
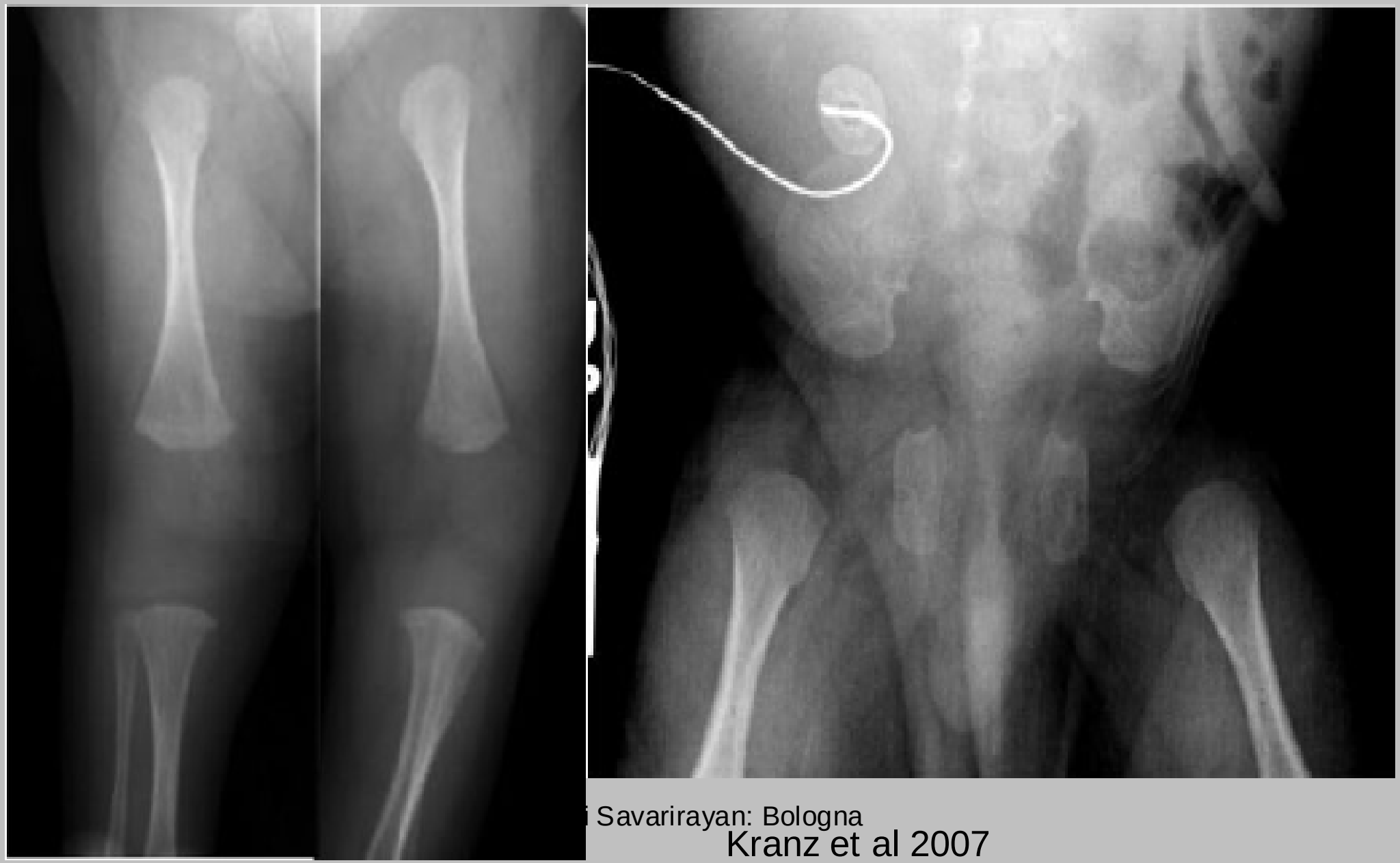


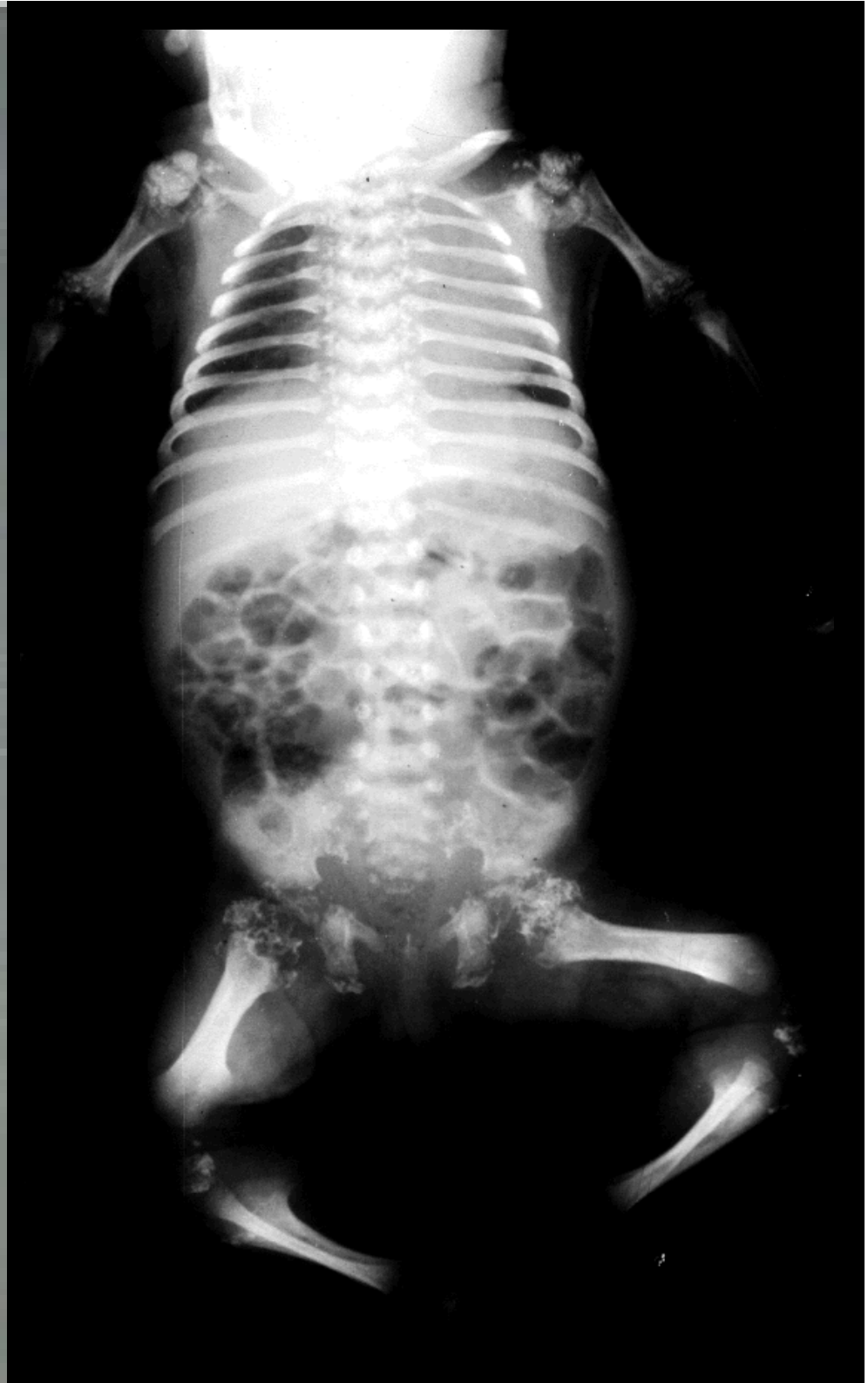
Sun et al 2005

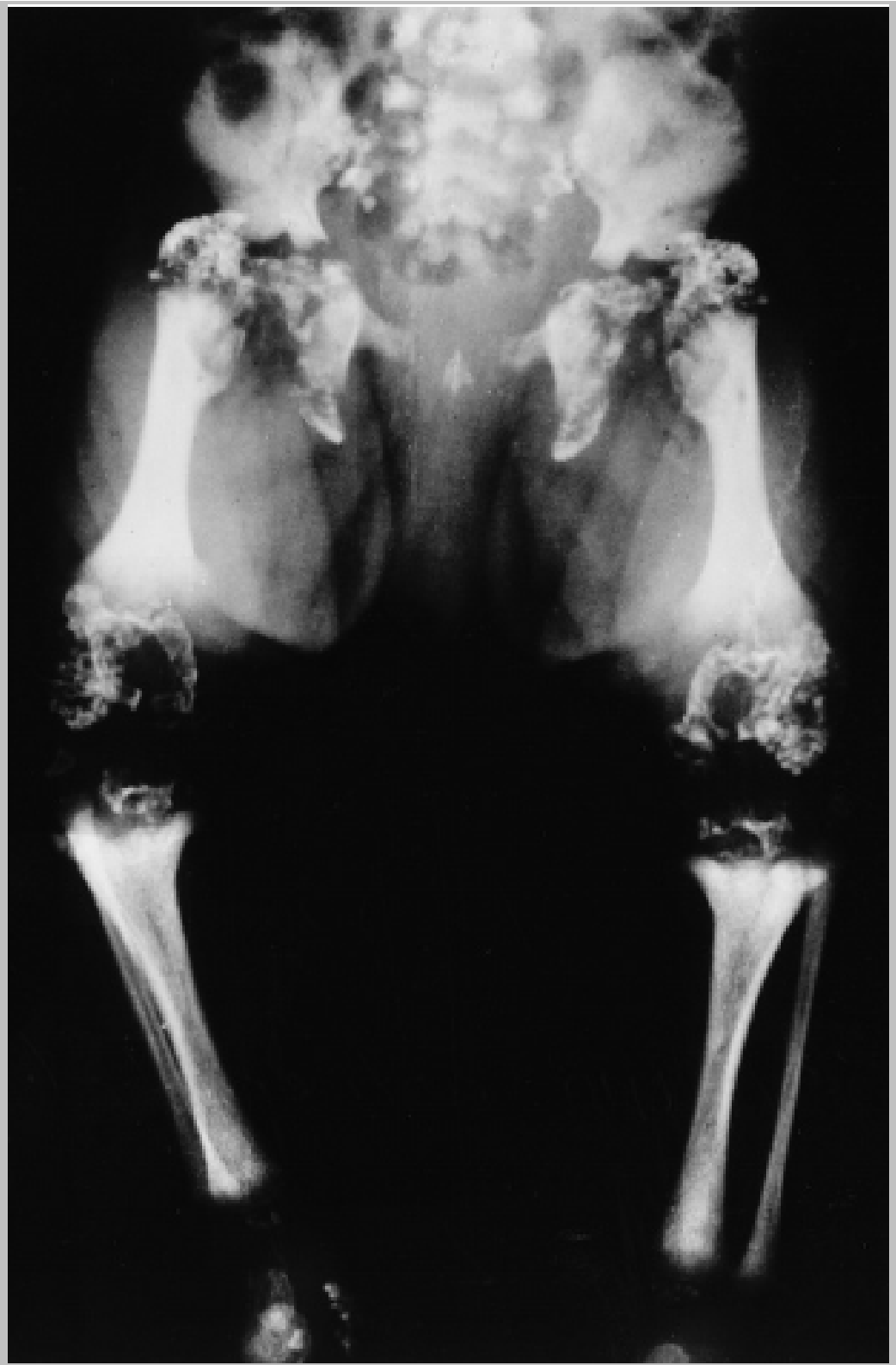
CDG-Ig



Kranz et al 2007

Peroxisomal Defects





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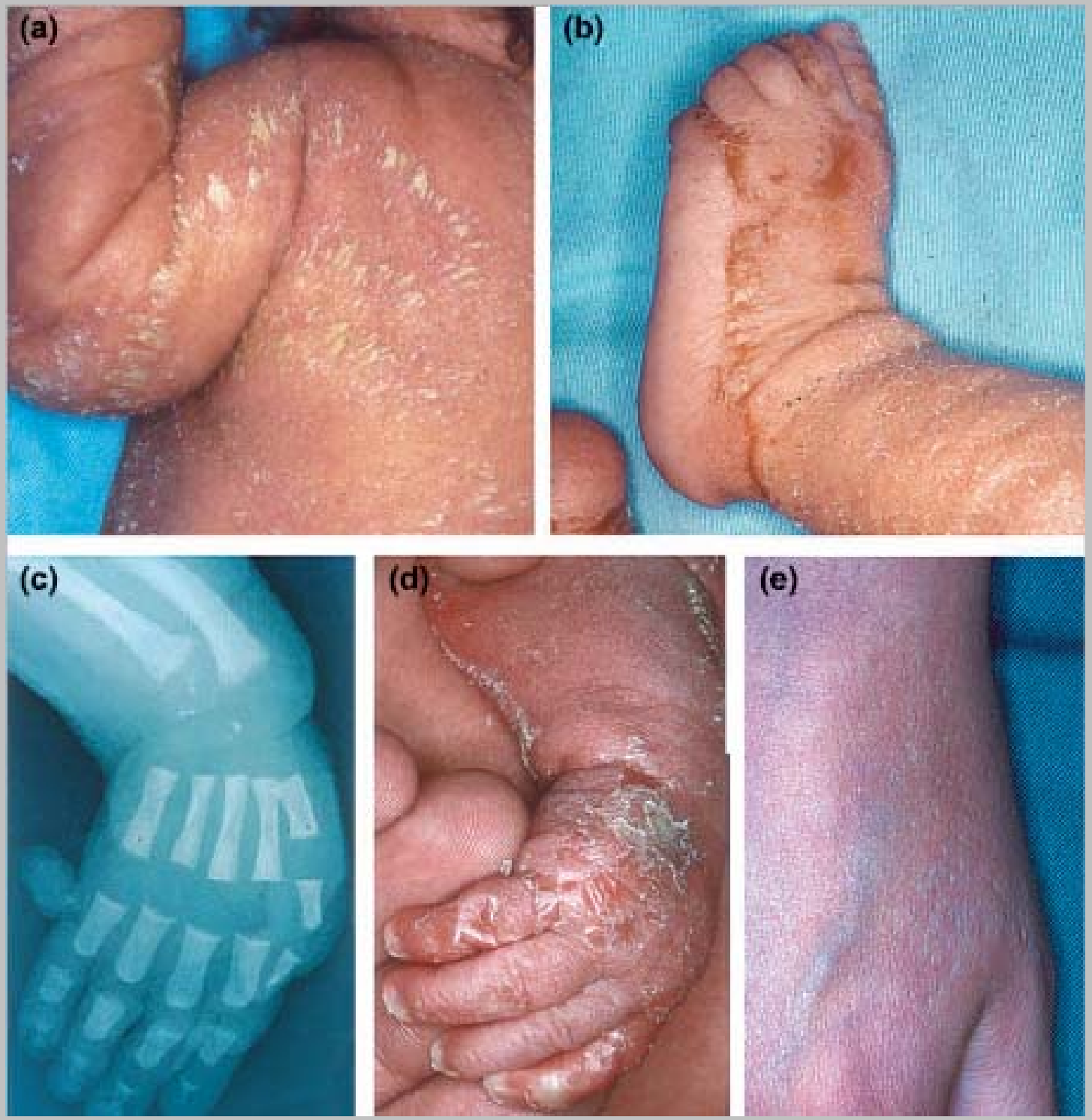
Cholesterol Biosynthesis Defects

X-CDP



Feldmeyer et al 2006

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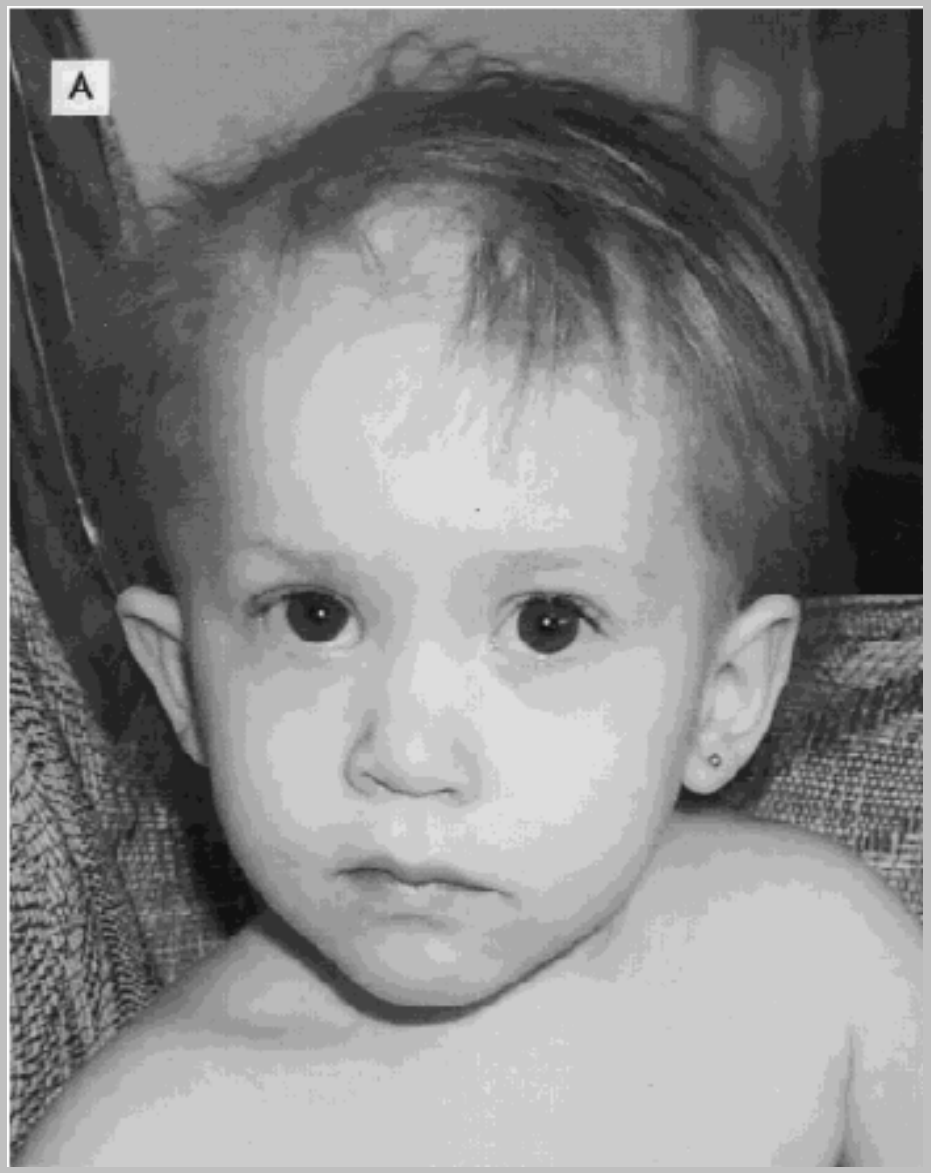
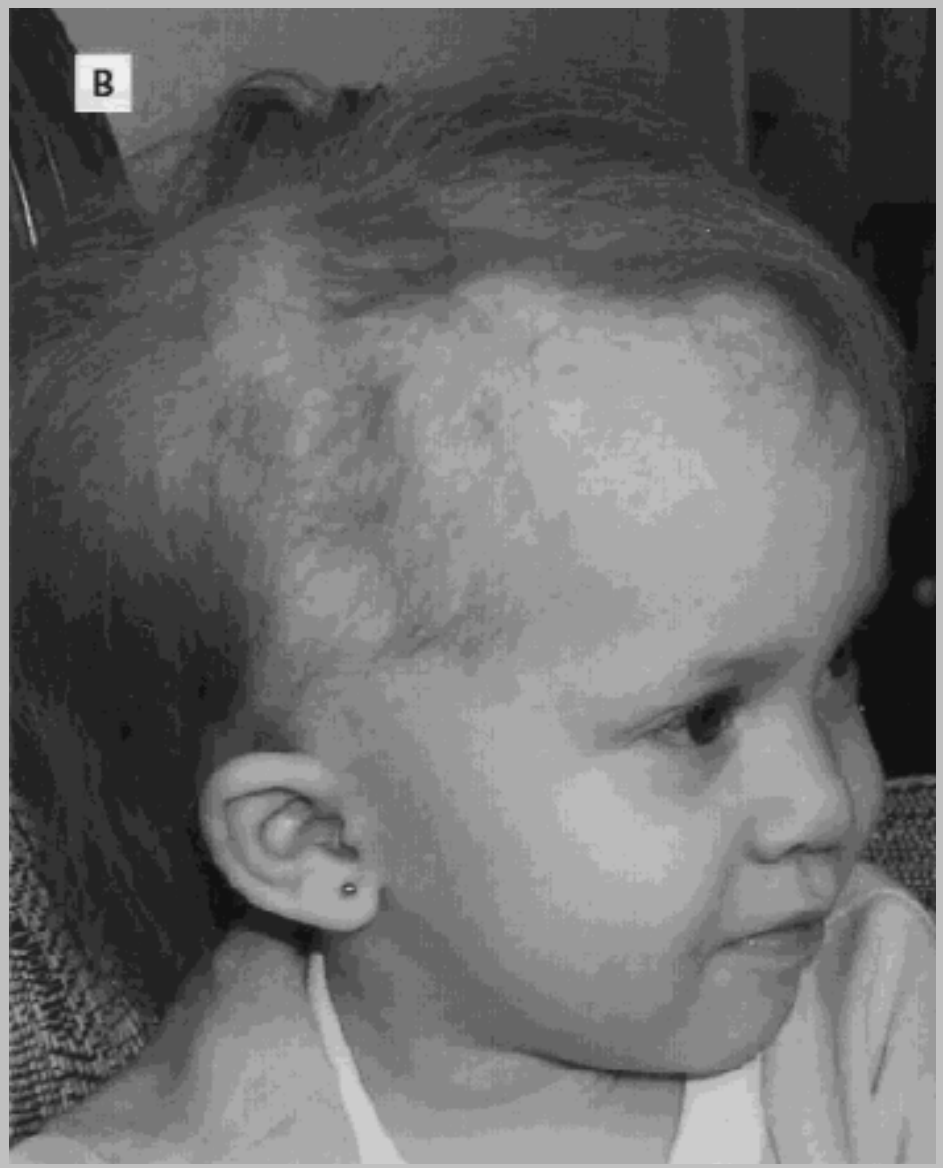
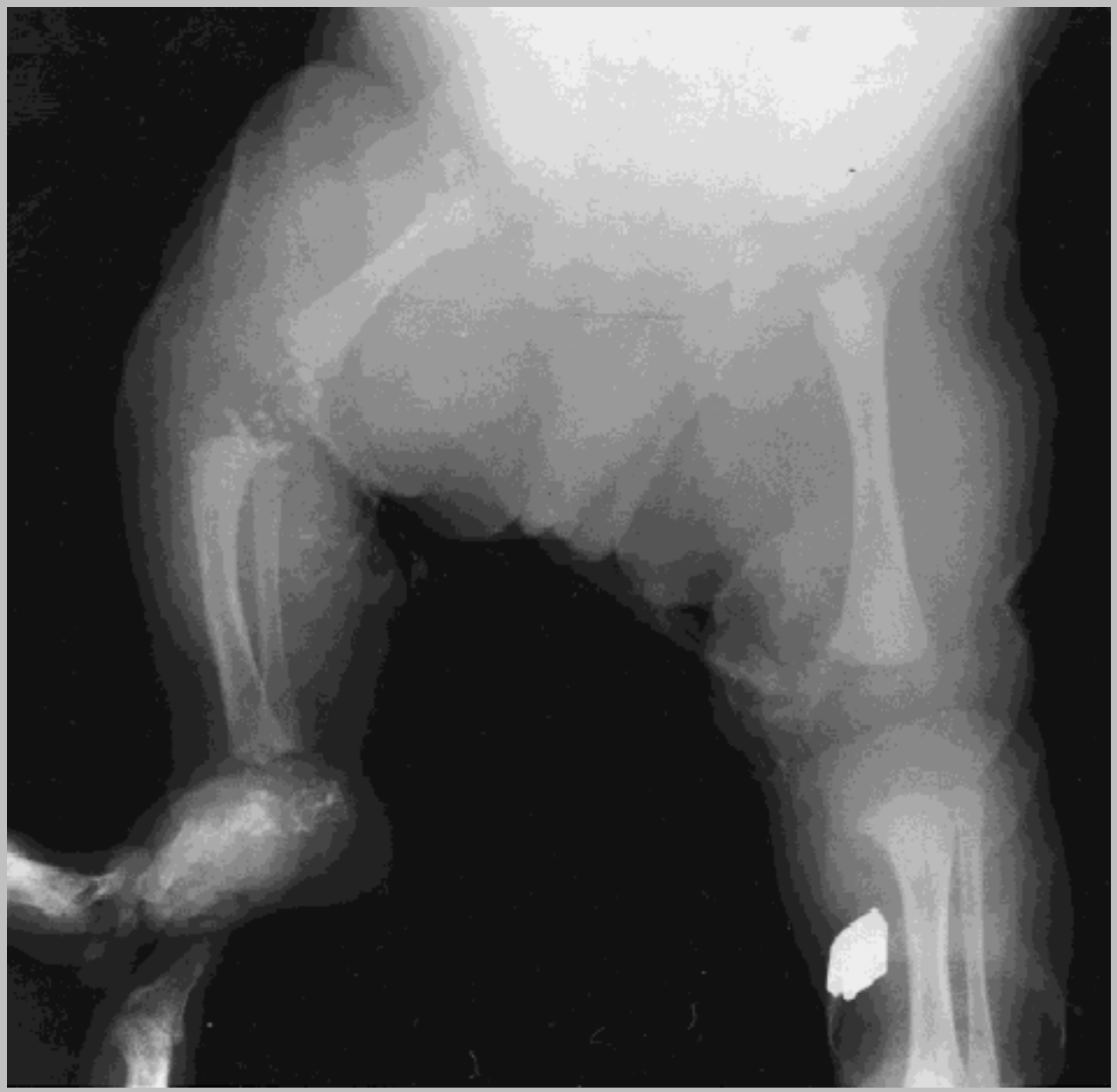
Congenital ***H***emidysplasia with ***I***chthyosiform naevus and ***L***imb deficiencies

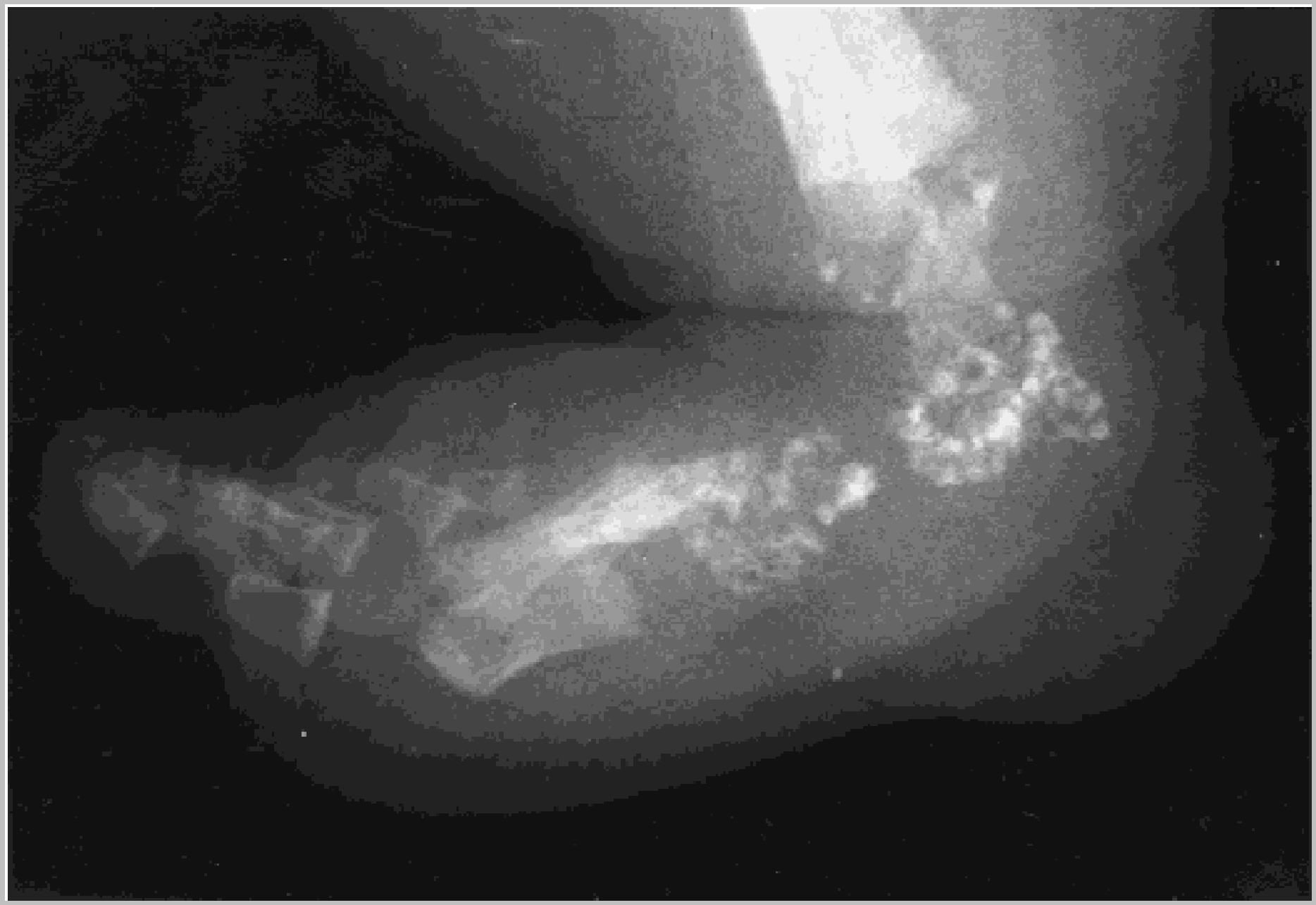




May 2008 Grange et al 2000 Ravi Savarirayan: Bologna



Grange et al 2000



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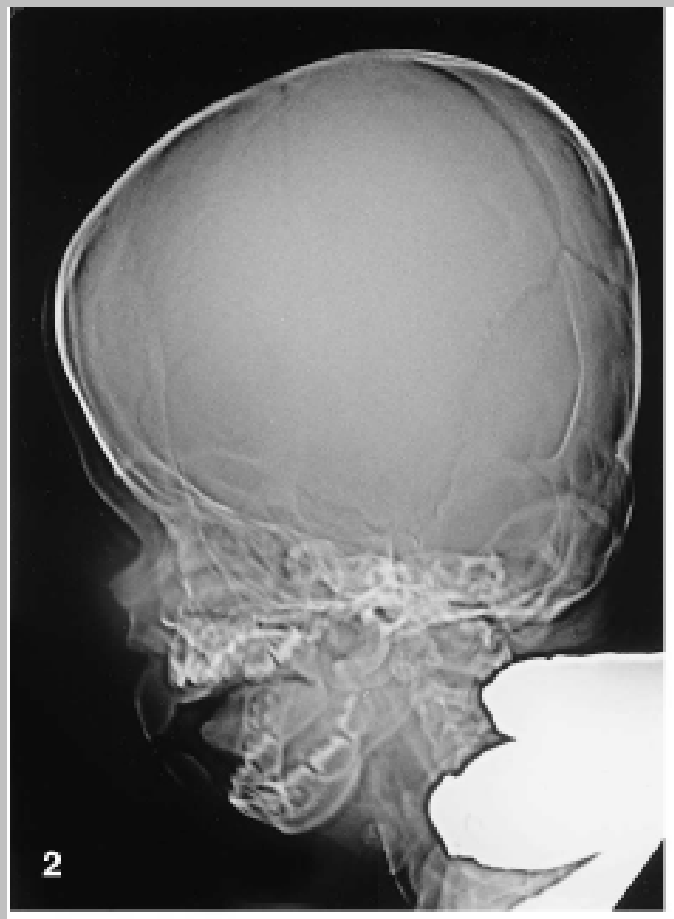
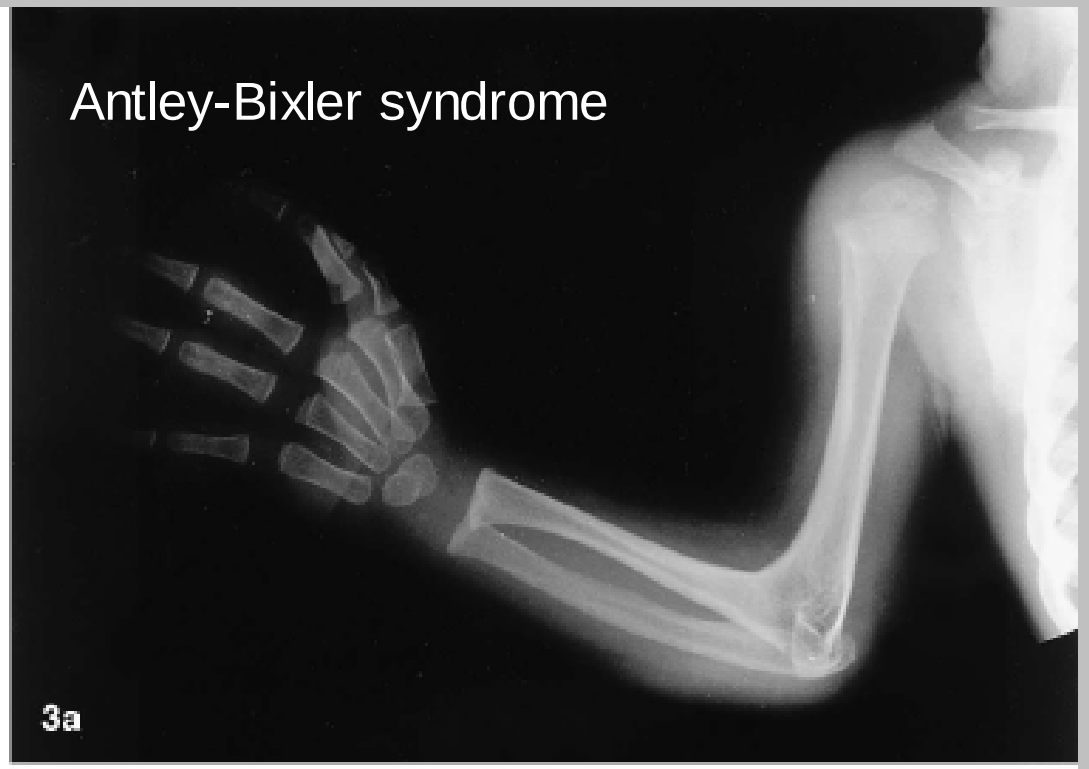
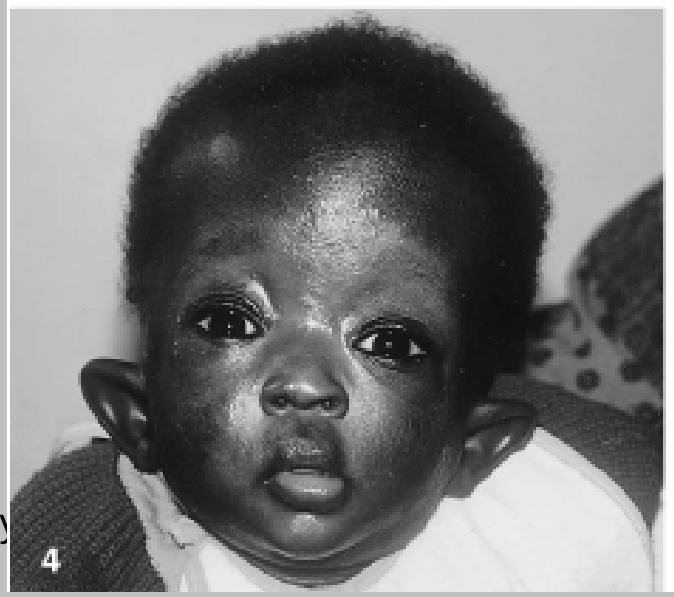
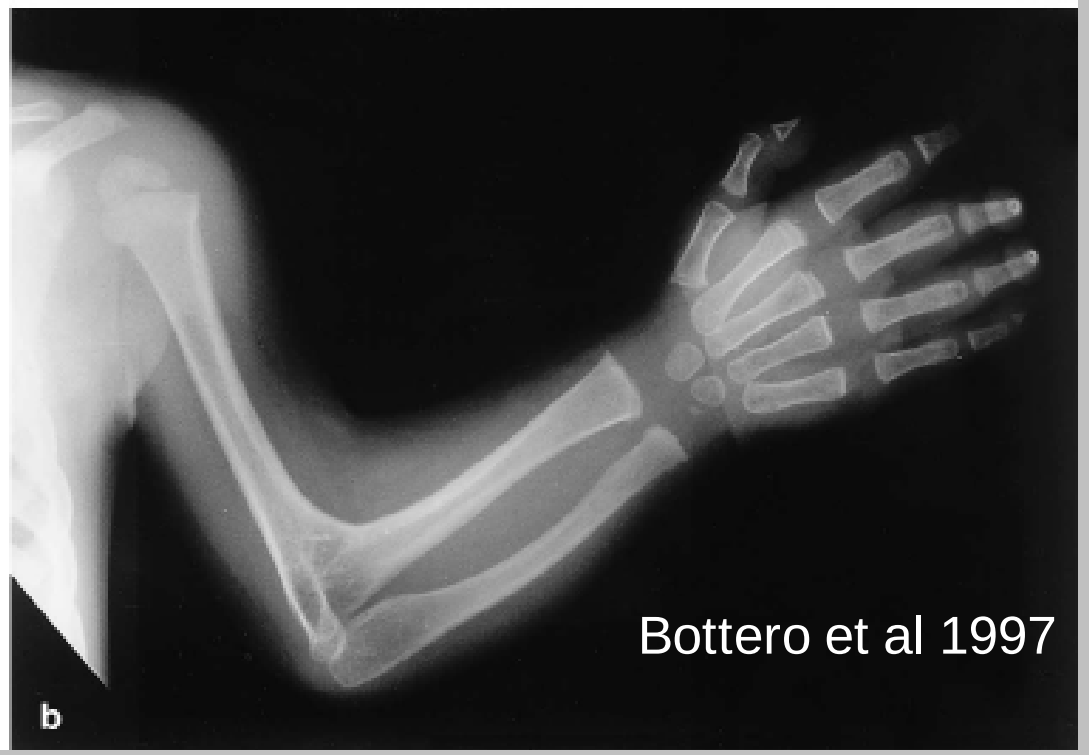
Grange et al 2000









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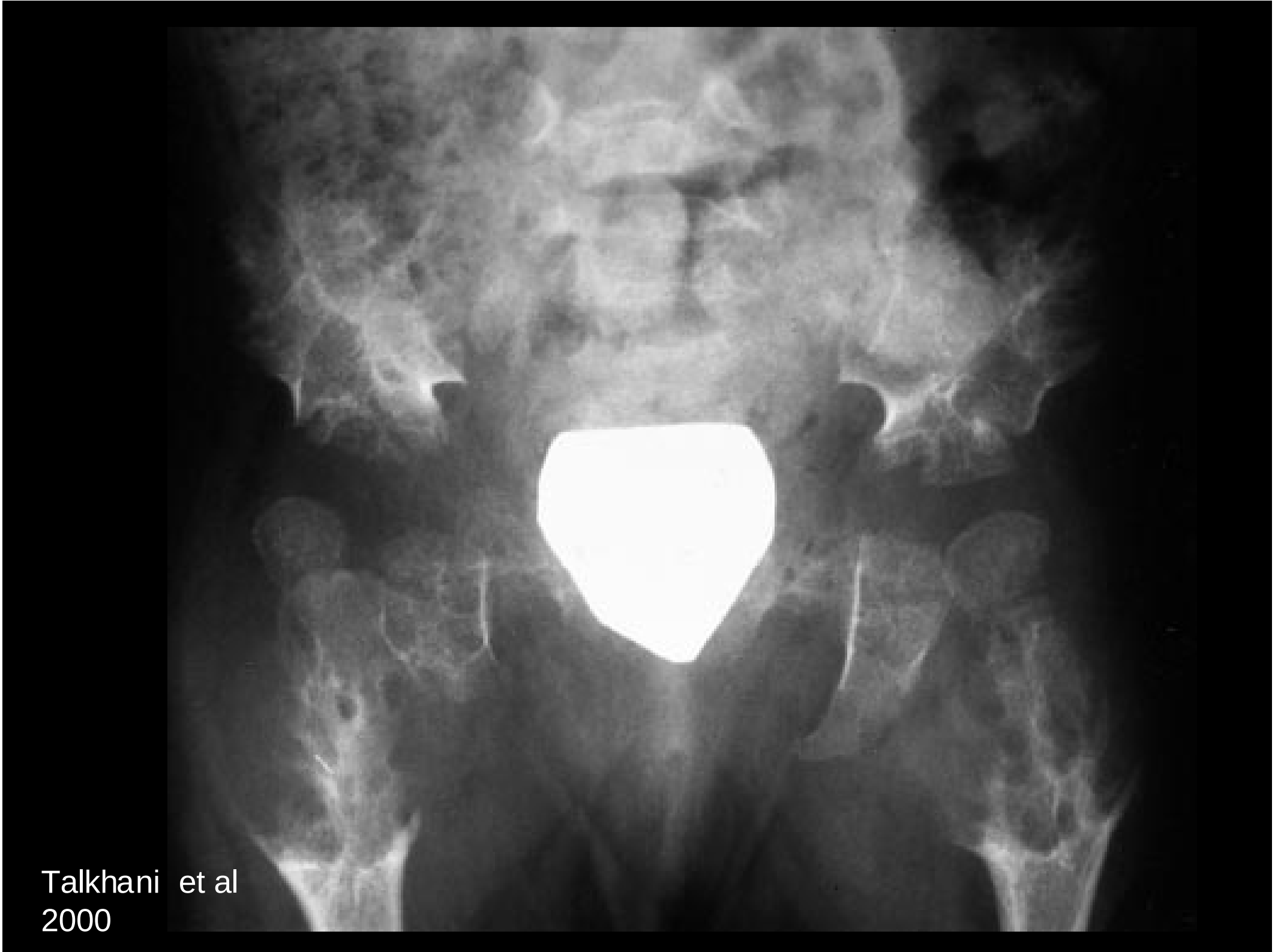
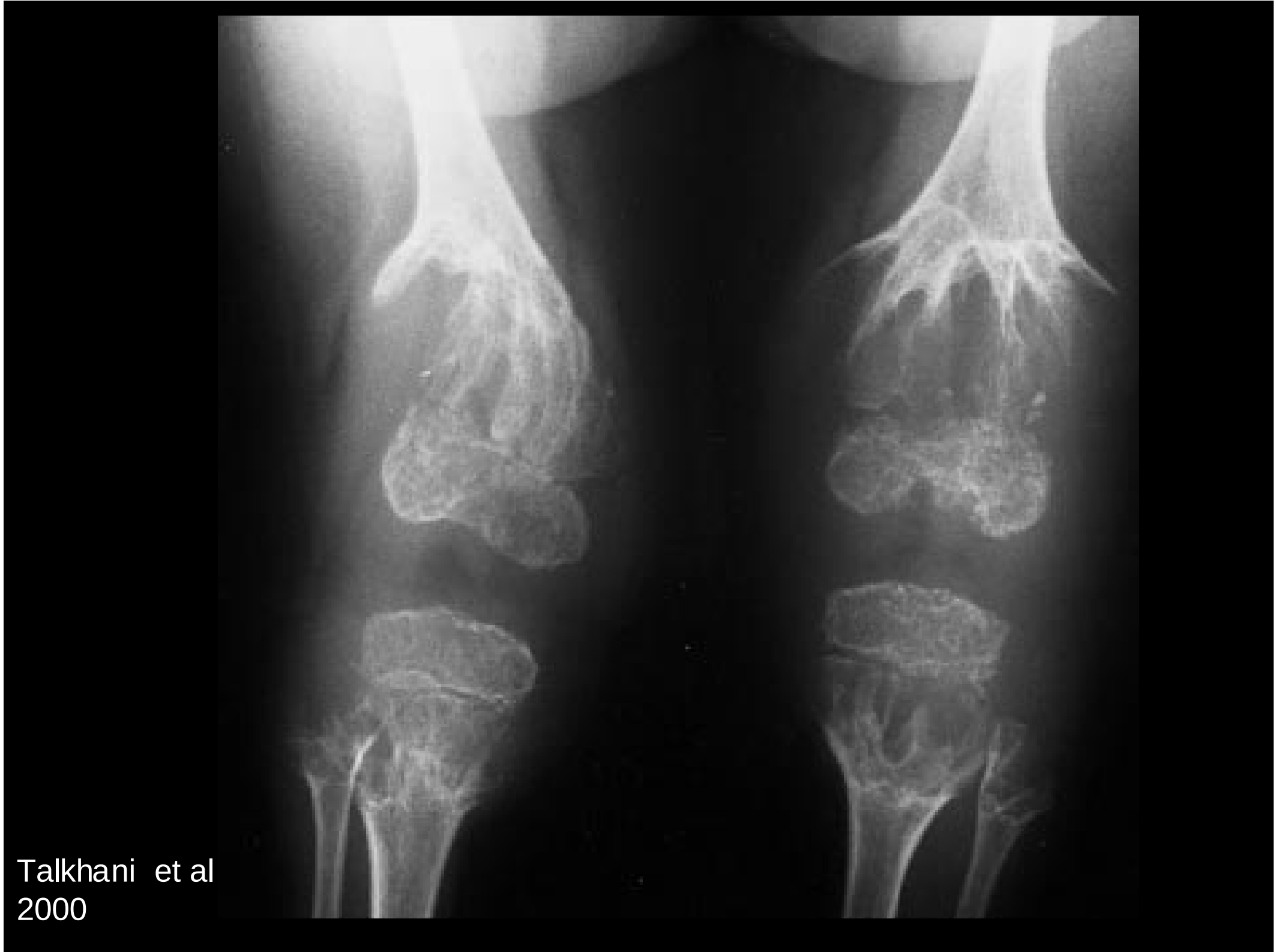
Others conditions

- Desmosterolosis
- SLO

2-D-hydoxyglutaric aciduria



Talkhani et al
2000



Talkhani et al
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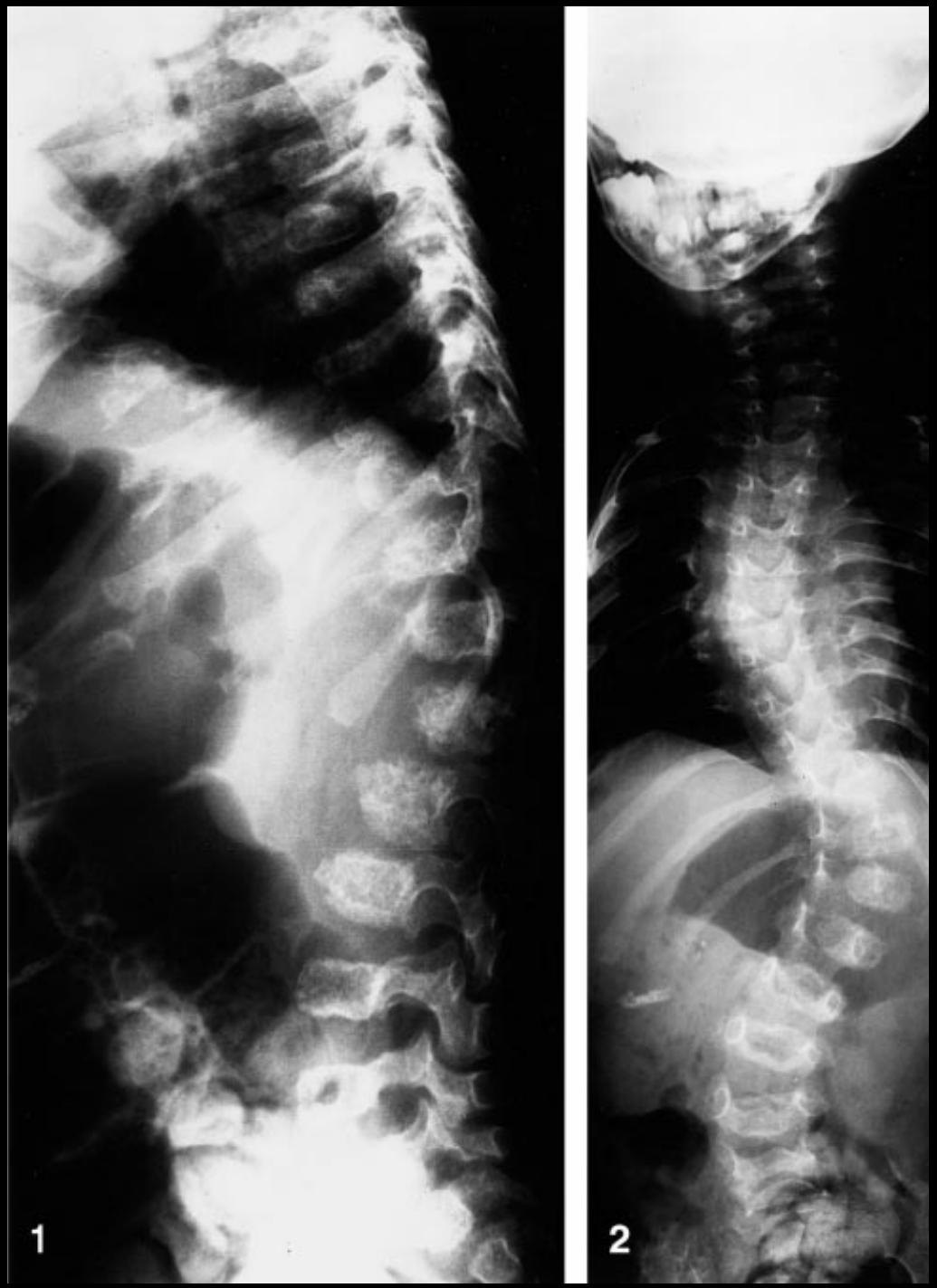

Talkhani et al
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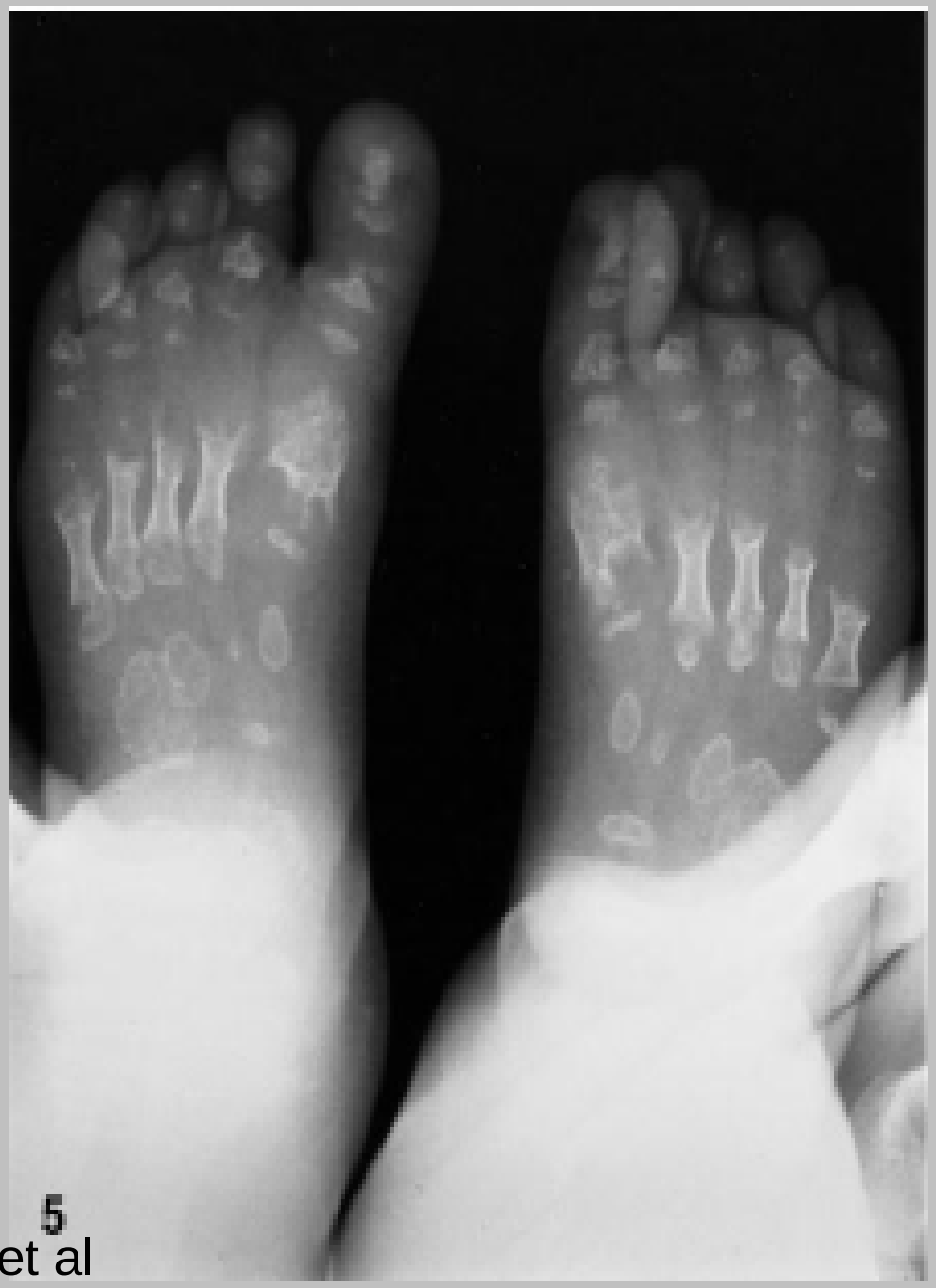
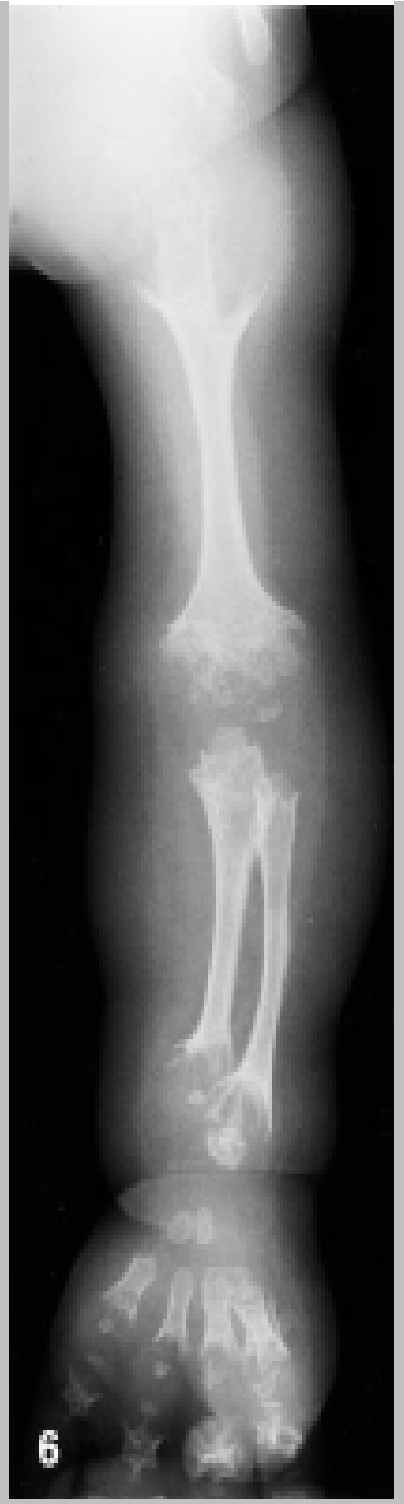


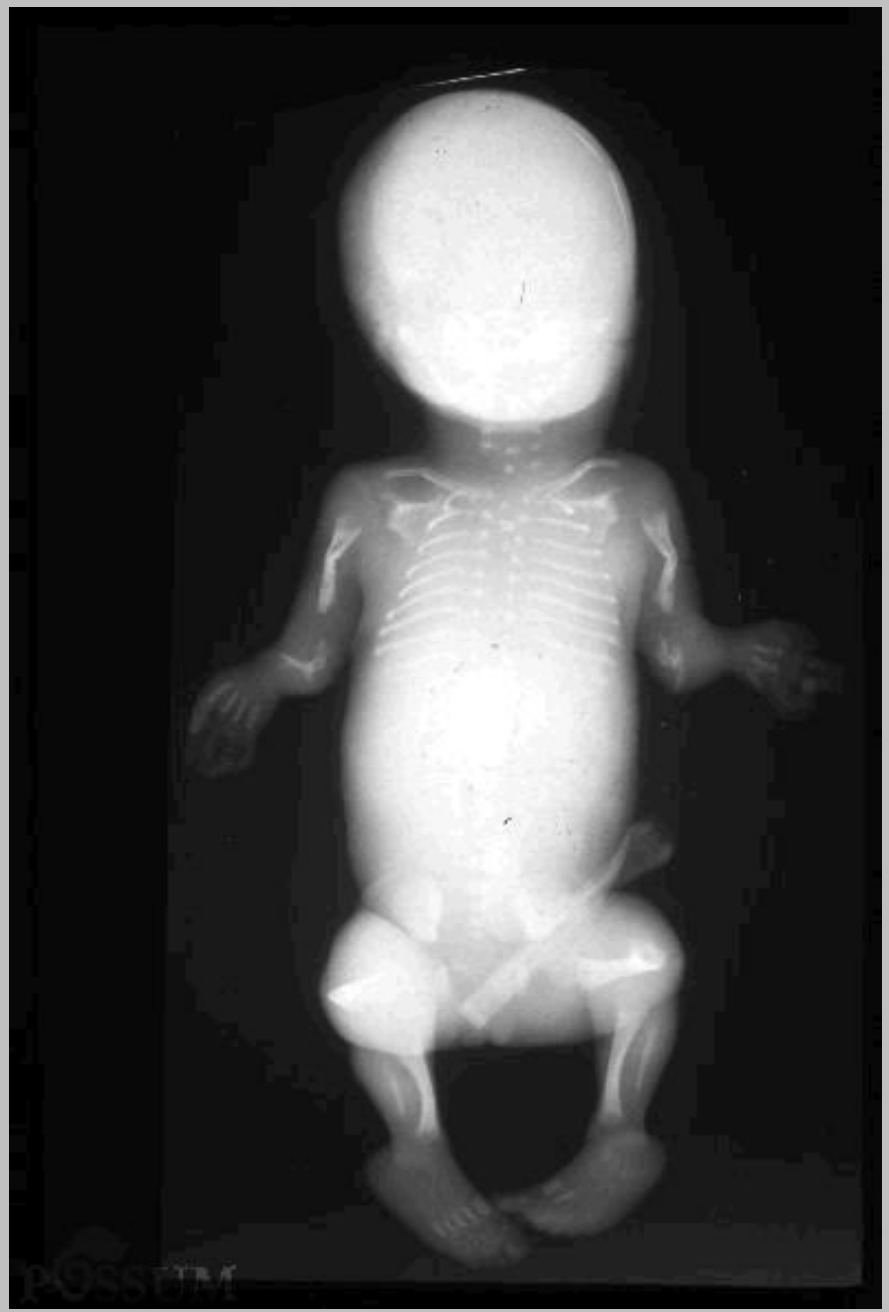


Talkhani et al 2000

IEM and Osteoporosis

- Homocystinuria
- Galactosaemia
- PKU
- Glycogen Storage Disease Type Ia
- Congenital Disorder of Glycosylation type Ia
- Lysinuric Protein Intolerance

Hypophosphatasia



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References

- *Coman and Savarirayan Clin Genet. 2008 May 6. [review]*
- *Coman and Savarirayan Am J Med Genet A. 2008 Feb 1;146(3):389-92.*