Short Rib Syndrome

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History
Stillborn male with anomalies.

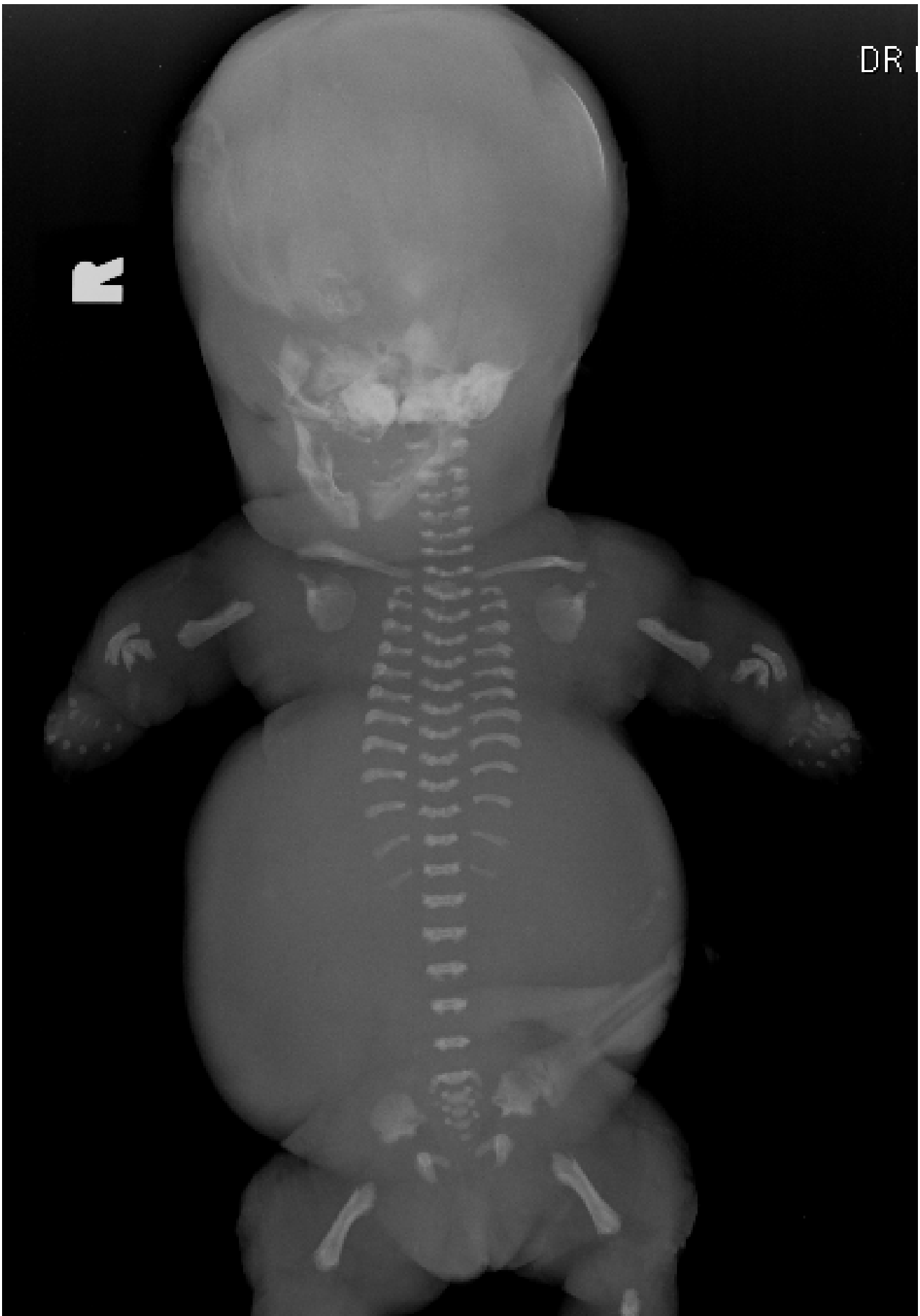
Diagnosis
Short Rib Syndrome

Additional Clinical
Autopsy-Significant findings include: 1) proboscis with absent nasal cavity, 2) single orbit containing 2 malformed oculi, 3) semilobar holoprosencephaly, 4) common AV valve with truncus malformation, 5) bowel malrotation, 6) pulmonary hypoplasia, 7) small hyperlobulated kidneys, 8) gonadal hypoplasia, 9) absent adrenal glands, 10) pharyngeal dysmorphism with cleft and 11) cartilage dysplasia.

Discussion
Short rib syndrome is an autosomal recessive lethal dysplasia. There are multiple types with significant radiological and pathological overlap. The above case demonstrates most of the significant radiographic findings.

Findings
CR-Significant findings include: 1) Anasarca, 2) short horizontal ribs with thoracic hypoplasia, 3) small round scapulae, 4) small ilia, 5) marked symmetric micromelia with very short tubular bones mesomelic greater than rhizomelic, 6) bowed radii and ulnae, 7) platyspondyly, 8) relatively large skull with thin calvaria, 9) flat mandibular angle, and 10) postaxial polydactyly with symphalangism.

Reference
Spranger JW, Brill PW, Poznanski A. Bone Dysplasias: An Atlas of Genetic Disorders of Skeletal Development 2nd Ed (2002).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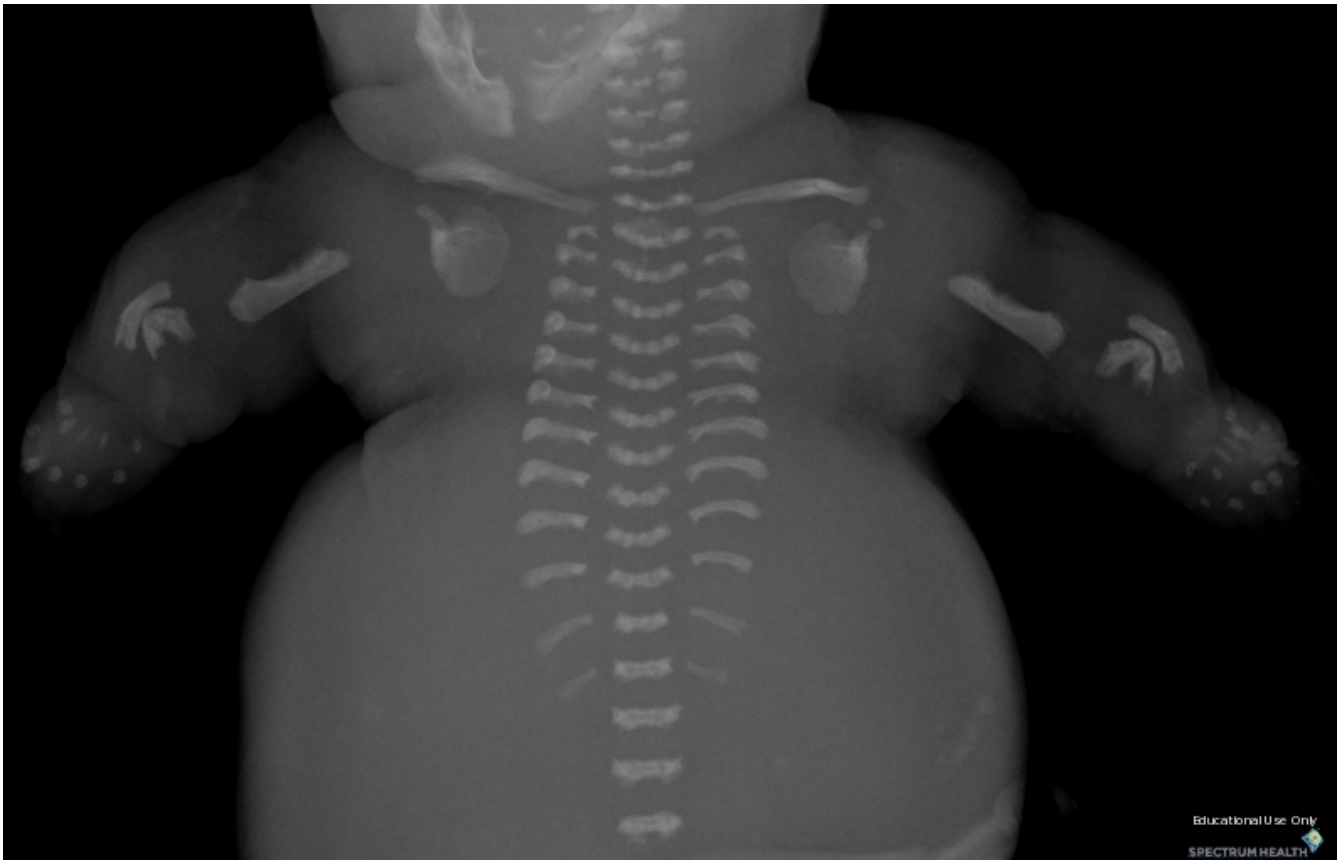
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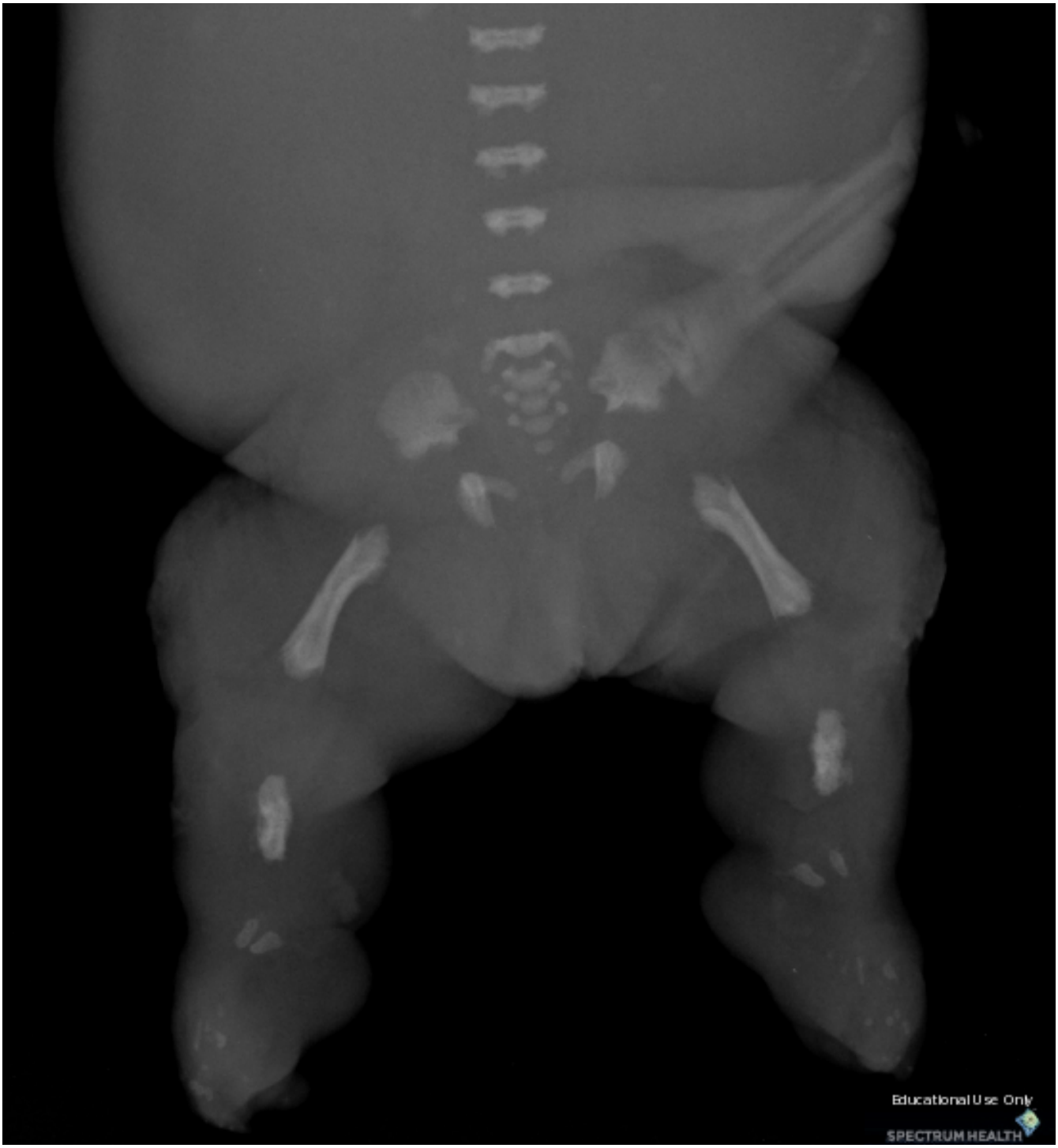
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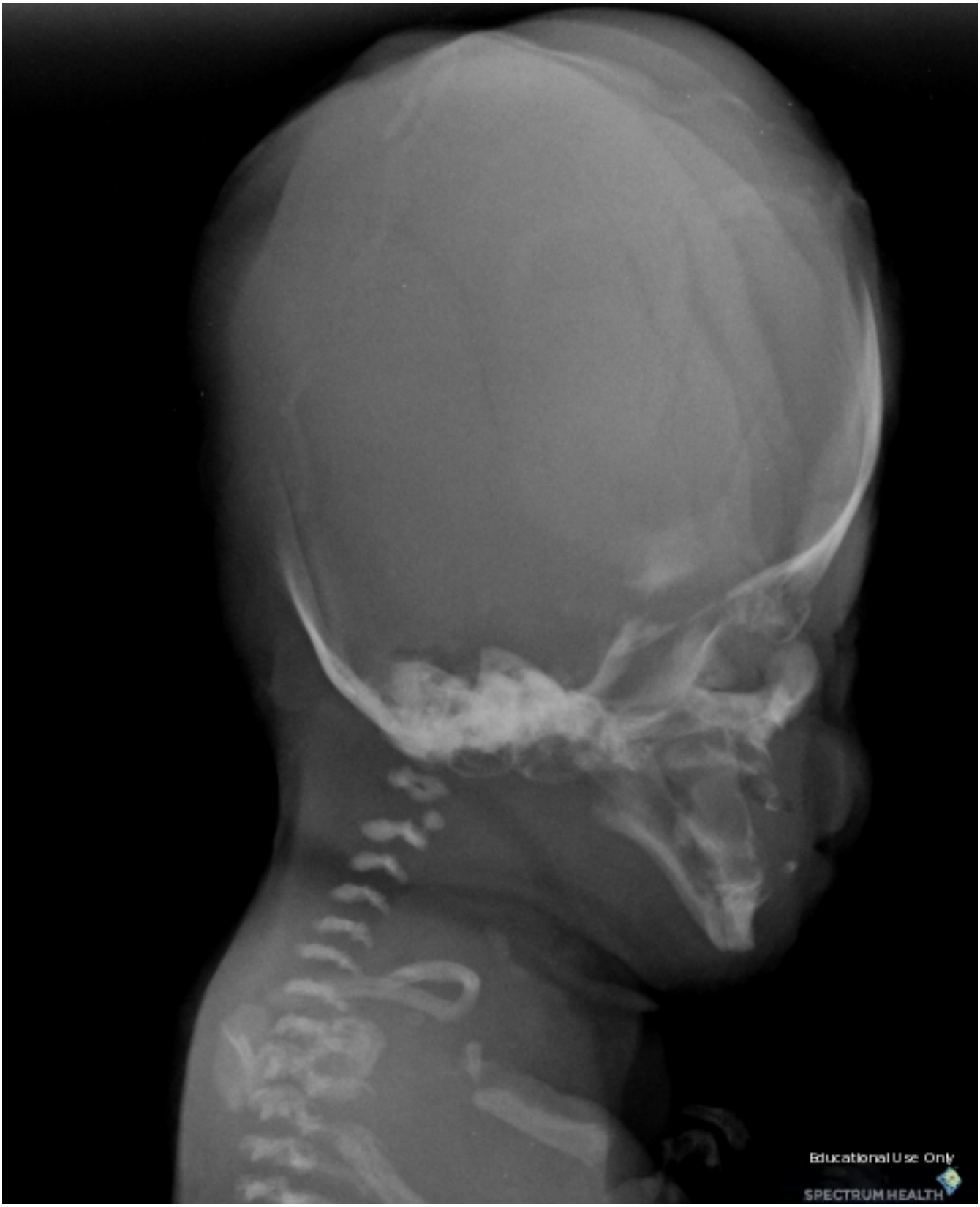
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