Short rib polydactyly syndrome

type III (Verma-Naumoff type)

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Section: Paediatric radiology
Case Type: Clinical Cases

Clinical History:

Full-term neonate presented for post-mortem. An ultrasound scan during pregnancy had shown a narrow chest and short limbs. Post-mortem x-ray examination was performed.

Imaging Findings:

A full-term baby (death 1 hour after delivery) was presented for post-mortem. The 19-year-old mother had concealed the pregnancy until 32 weeks. Ultrasound at presentation in pregnancy had shown a narrow chest and short limbs, but definite diagnosis was not made at this stage. Caesarean section was undertaken for maternal distress. A live-born fetus was delivered and died at 1 hour. The baby was noted to have an extremely narrow chest, pulmonary hypoplasia, simian creases and polydactyly. Post-mortem examination revealed shortening of limbs, especially proximally, extremely narrow chest, post-axial hexadactyly of hands and feet, normal male genitalia, hypoplastic lungs and kidneys, coarctation of the aorta and biventricular hypertrophy. Post-mortem x-ray examination was performed.

Discussion:

The short rib polydactyly syndromes (SRPS) are lethal forms of short-limbed dwarfism. Type III (Verma-Naumoff type) is similar to type I (Saldino-Noonan) but has less severe clinical and radiological features. Both types show
autosomal recessive inheritance. The underlying gene has not yet been discovered. Clinically, the foetal abnormalities include micromelic dwarfism, polydactyly of hands and feet, cleft lip/palate, and narrow thorax.

Radiologically, there is rhizomelia with metaphyseal spikes of the long bones, giving a "ball in cone" appearance. The hands and feet are well ossified with post-axial polydactyly in both hands and feet, although this may not always be the case. The scapulae have an irregular outline, and the thorax is narrow, with short ribs. The vertebrae are reduced in height and have irregular contours with increased disc space. The iliac bones are triangular and small, with prominent spikes at the sciatic notch. The skull vault is well ossified, with frontal bulging and a flattened occiput. Other reported abnormalities include renal, gastrointestinal, cardiovascular and genital anomalies.

Differential diagnoses include other SRPS, particularly type I; Ellis-van Creveld syndrome; and asphyxiating thoracic dysplasia. Distinguishing features of Type I SRPS from Type III SRPS include the absence of fibulas, a female preponderance, pointed femoral ends and more urological and cardiovascular abnormalities.

The risk of recurrence of this condition in another pregnancy is 25%. There are no specific biochemical or histopathological markers for SRPS and therefore early ultrasound is a valuable tool for identification.

Differential Diagnosis List: Short rib polydactyly syndrome Type III (Verma-Naumoff type)

Final Diagnosis: Short rib polydactyly syndrome Type III (Verma-Naumoff type)

References:

Description: There are tubular long bones with a ball-in-cone appearance at the metaphyses. The vertebrae are decreased in height with increased disc space. Origin:
**Figure 2 a**

**Description:** There are broad irregular scapulae and a narrow thorax with short, broad ribs. There is post-axial hexadactyly of the hands. The skull vault is well ossified. **Origin:**