Dysmorphology report

Unknown syndrome: radial ray defects, omphalocele, diaphragmatic hernia, and hepatic cyst

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Abstract
We present a male infant with a giant omphalocele, diaphragmatic hernia, hepatic cyst, bilateral radio-ulnar synostosis, absent left thumb, and triphalangeal right thumb.

History
Prenatal. Polyhydramnios. Giant omphalocele shown by ultrasonography.

Birth. Caesarean section because of double footling breech presentation. Apgar scores 4 and 8 at one and five minutes.

Family. First child of healthy, unrelated parents. Father aged 34 years, mother aged 27 years.

Medical history
At birth, primary repair of the giant omphalocele failed. A prosthetic silo inserted into the abdominal wall, followed by gradual reduction of contents. Reoperated at 10 days. Intrahepatic cyst containing clear fluid unroofed. Peritoneal cavity filled with lymphatic fluid draining into the right chest through a right posterior diaphragmatic hernia. Hernia repaired and abdominal wall closed. Died two days after operation. Permission for necropsy not granted.

Clinical examination
Birth weight 3000 g (25th centile), length 50 cm (25th centile), head circumference 35.5 cm (50th centile). Face unusual, short, downward slanting palpebral fissures, bushy eyebrows, high nasal bridge, short

Figure 1 Patient's face and general appearance.

Investigations
Normal G banded male karyotype. X rays showed bilateral proximal radioulnar synostosis, absence of right metacarpal and phalanges, and three normal first phalanges. Normal radiographs of feet, spine, and skull. Ultrasonography of the abdomen showed large cystic lesion in a midline liver.

Discussion
The patient had many facial and acral manifestations typical of Nager syndrome (NS), including short, downward slanting palpebral fissures, high nasal bridge, short nose, small, upturned nostrils, malar and mandibular hypoplasia, dysplastic ear, short neck with low hairline, and tongue-like extension of hair on both cheeks. Other findings, such as hirsutism, bushy eyebrows, a long, thin upper lip with a midline beak, and thick skin, are characteristics of Brachmann-de Lange syndrome (BDLS), but normal birth weight, length, and occipitofrontal circumference and preaxial limb deficiencies are not. The radial ray defects in our patient were very similar to those reported in NS. However, he did not show cleft palate or absence of lashes, and the ears were less dysmorphic and the micrognathia less severe than usually seen in NS. An omphalocele, a diaphragmatic hernia, and a hepatic cyst have not been reported in either NS or BDLS. A diaphragmatic hernia and an omphalocele have recently been described in three patients with Fryns syndrome, which is characterised by distal digital hypoplasia and is not applicable to our case.

The mode of inheritance of this new syndrome is unknown. It may be sporadic, but autosomal recessive inheritance cannot be ruled out.

Figure 2. (a) Left hand with absent thumb. (b) Right hand with triphalangeal finger-like thumb.

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