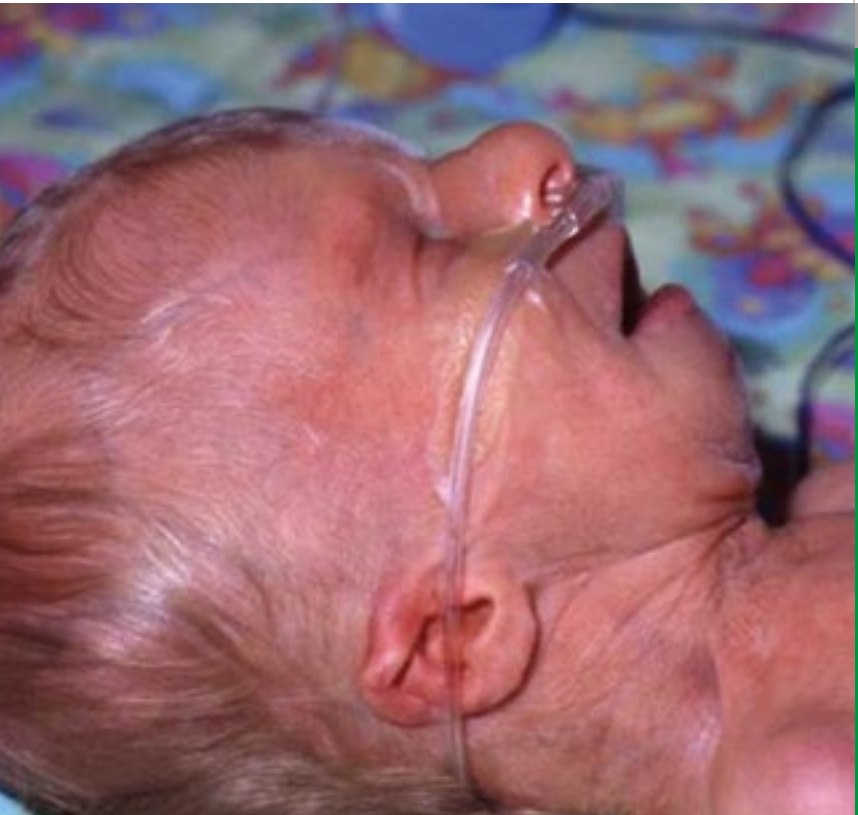
SWISS SOCIETY OF NEONATOLOGY

Yunis-Varon syndrome



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INRODUCTION

We report a newborn boy with the very rare Yunis-Varon syndrome. An undefined dysmorphic syndrom was detected at 29th week of gestation by prenatal ultrasound. The syndrome could not be determined and, due to expected poor prognosis, consent was given by the parents to withhold intensive care after birth. However, the child survived and this caused intense grief, especially feelings of guilt and doubts about the perinatal management.

CASE REPORT

The boy was the second child of healthy, non-consanguineous Swiss parents whose first child, a girl, was normal. He was delivered vaginally at 37 3/7 weeks of gestation. Three previous pregnancies had ended in spontaneous abortions, and in one aborted fetus trisomy 7 had been detected. Otherwise family history was unremarkable.

The initial course of pregnancy had been uneventful. There was no evidence of intrauterine infection or maternal drug abuse. Prenatal ultrasound at 29 weeks of gestation showed intrauterine growth restriction, oligohydramnion and multiple fetal dysmorphic features. Amniocentesis revealed a normal karyotype (46XY).

After delivery, the patient was bradycardic and cyanotic, but showed spontaneous breathing efforts. With supplemental oxygen, his clinical condition impro-

ved. Apgar scores were 5, 6 and 6 at 1, 5 and 10 minutes, respectively, and umbilical pH values were 7.43 (arterial) and 7.51 (venous).

On admission, the boy was noted to be symmetrically growth restricted with a birth weight of 1520 g (700 g below P3), a length of 42.5 cm (2.5 cm below P3) and a head circumference of 29 cm (2.8 cm below P3).

There was an accentuated second heart sound with a 2/6 systolo-diastolic murmur, diastasis of the rectus abdominis muscle, an umbilical hernia measuring 5x5 cm (Fig. 1), an abnormally soft skull with wide sutures and confluent large anterior and posterior fontanels. Additional dysmorphic features were a high forehead, a low nasal bridge, a wide nose with anteverted nostrils, dysplastic, low-set ears and micro- and retrognathia (Fig. 2, 3).

Fingers and toes were short, pointed, with nail hypo-/aplasia and 50% skin syndactylies of fingers II/III and IV/V bilaterally (Fig. 4, 5). Halluces were very short and proximally implanted (Fig. 6, 7). The infant had general muscular hypotonia. Grasping and suckling reflexes were normal.

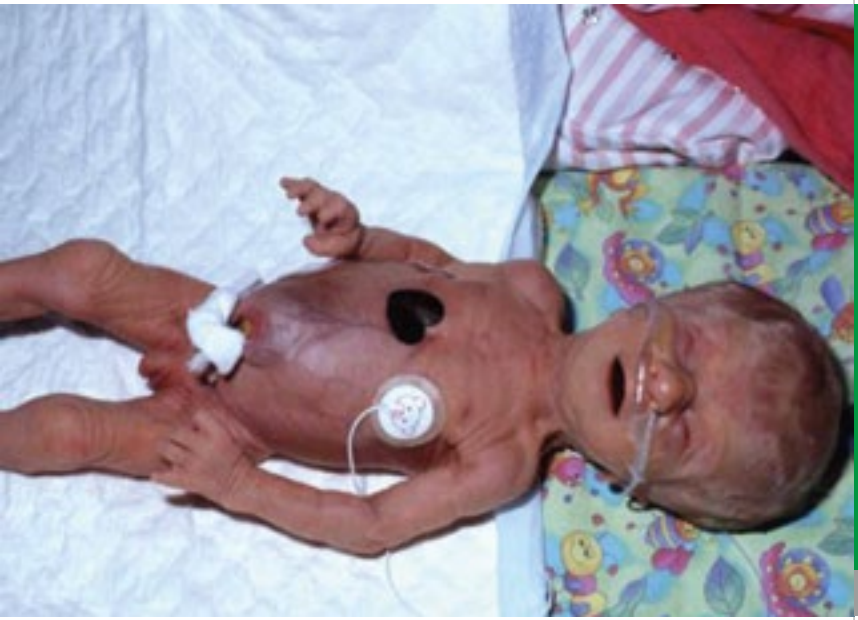

Fig. 1

Growth restriction.

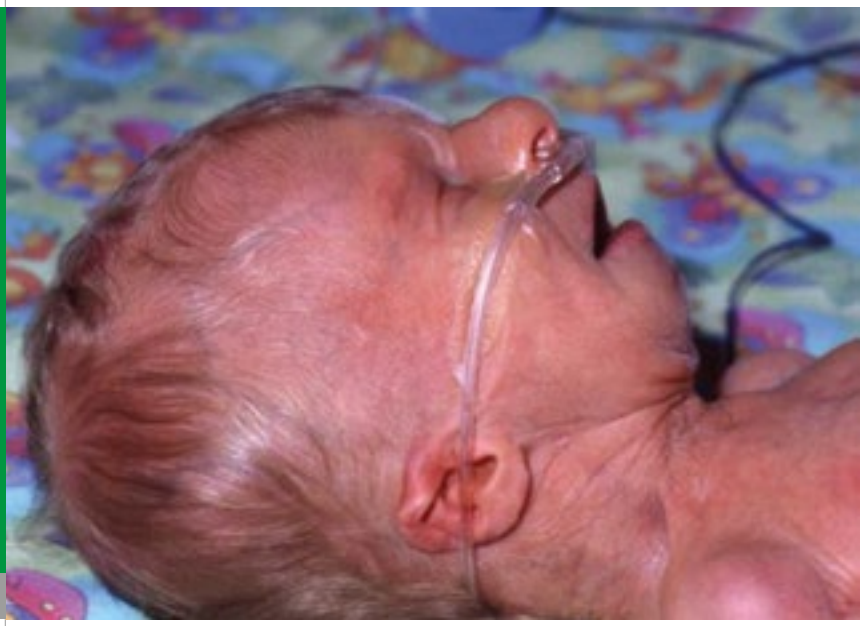

Fig. 2

Facial dismorphism.

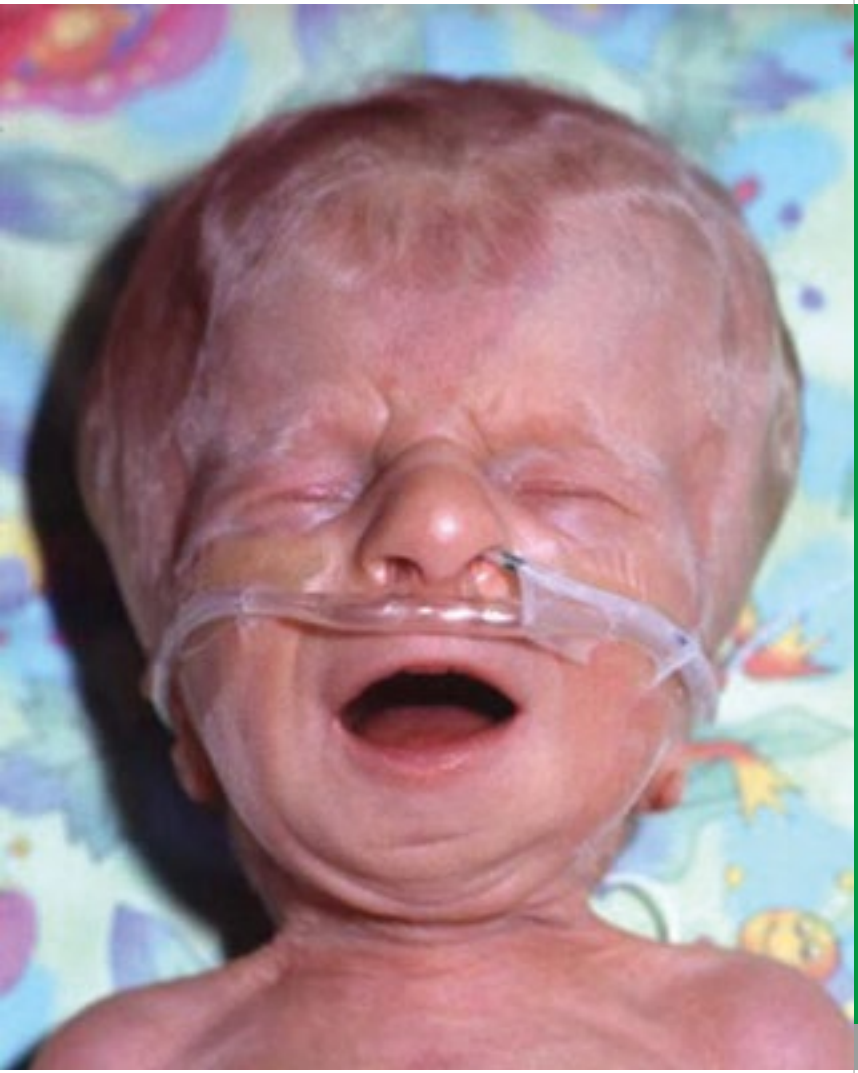

Fig. 3

Facial dismorphism.

Echocardiography showed a widely open ductus arteriosus Botalli with bidirectional shunt, tricuspid insufficiency and pulmonary hypertension. On abdominal sonography, there was no spleen and slight hepatomegaly, as well as right-sided hydronephrosis and hydroureter. Radiological findings included missing distal phalanges of hands and feet and severe osteopenia. Cranial sonography showed no abnormalities. Routine blood tests were normal except for the presence of Howell-Jolly bodies and slightly elevated values for creatinine and lactate. Urine analysis detected toluidin, fumarate and elevated glycin.

Based on these findings, the diagnosis of Yunis-Varon syndrome was made. The boy recovered from the respiratory distress syndrome within a few days. Nutrition was mainly managed by feeding through a nasogastric tube. At the age of 22 days, the patient was discharged at home in good general condition.

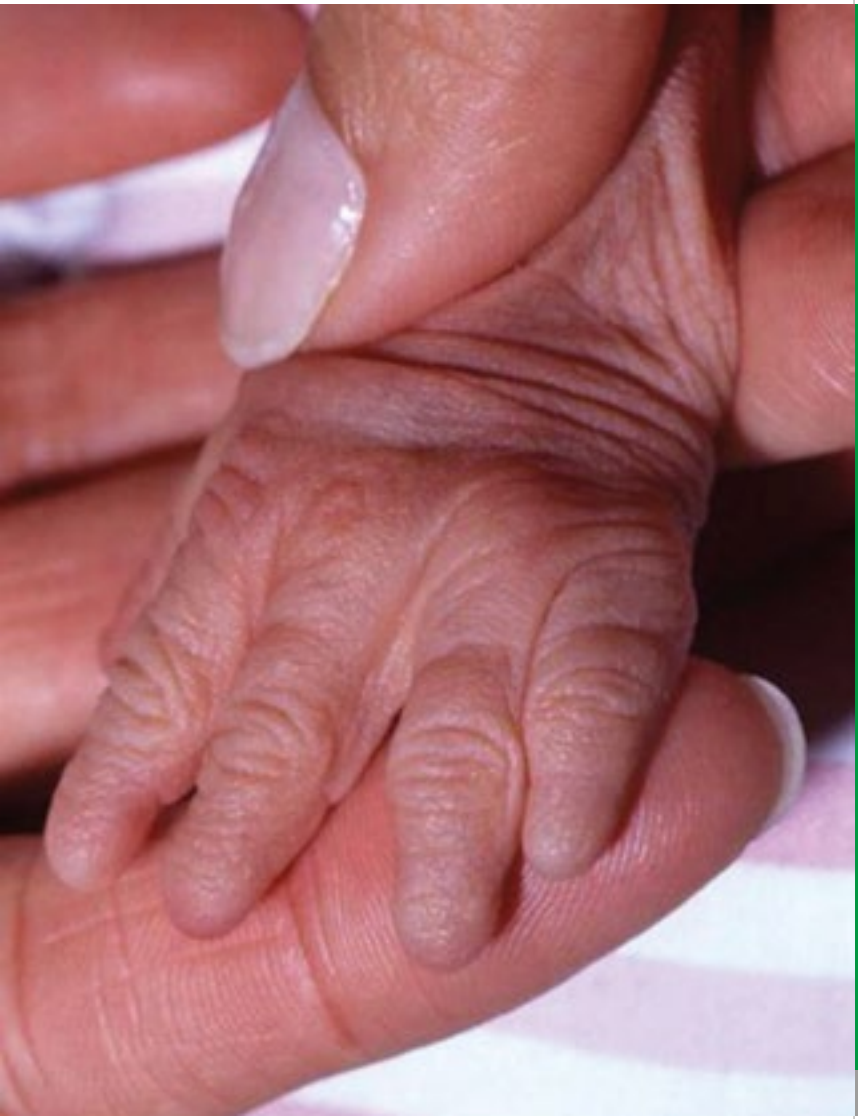

Fig. 4

Syndactylies and distal hypoplasiy of fingers.

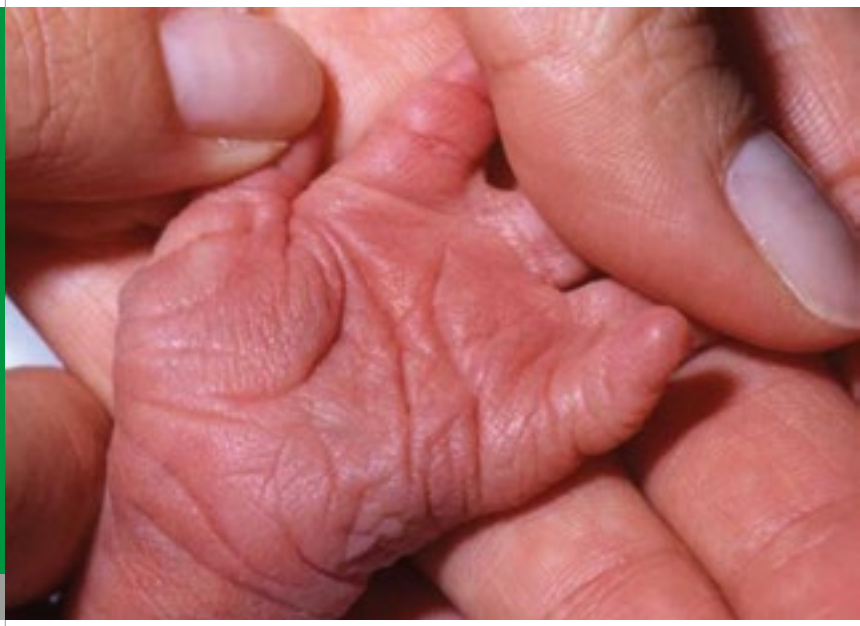

Fig. 5

Syndactylies and distal hypoplasia of fingers.

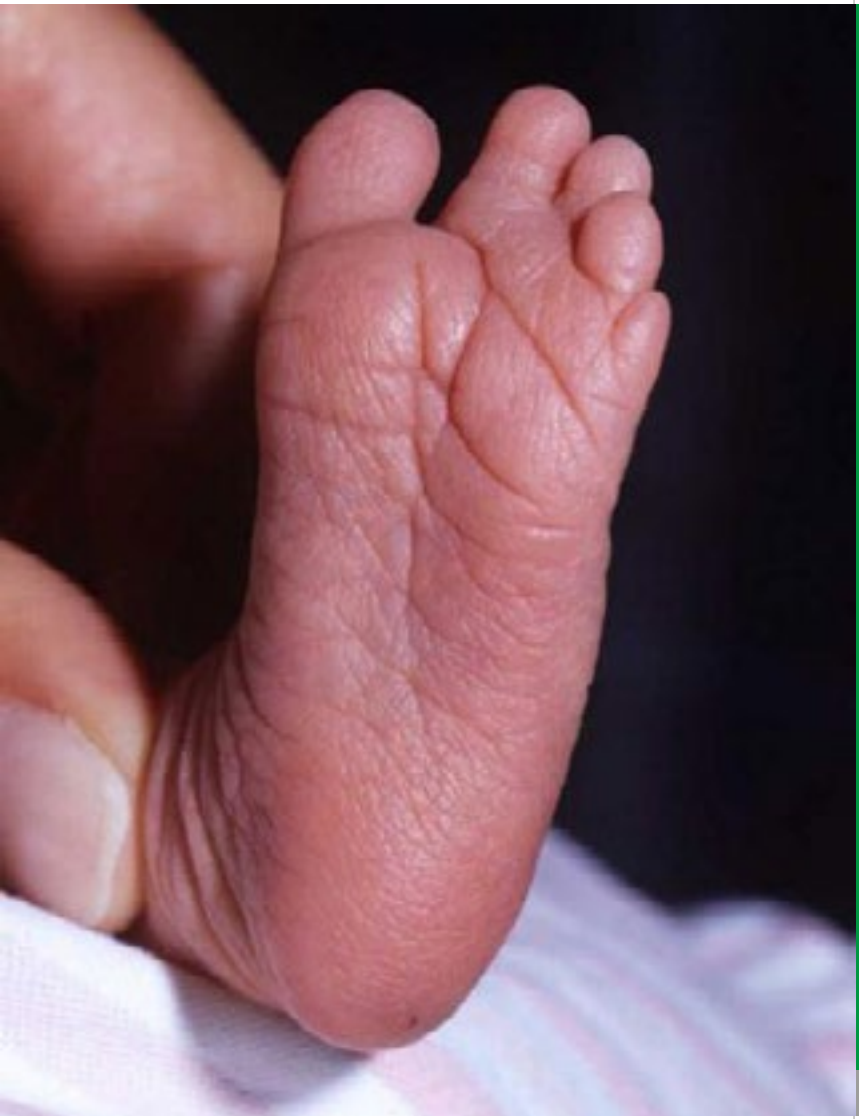

Fig. 6

Syndactylies and distal hypoplasia of toes.

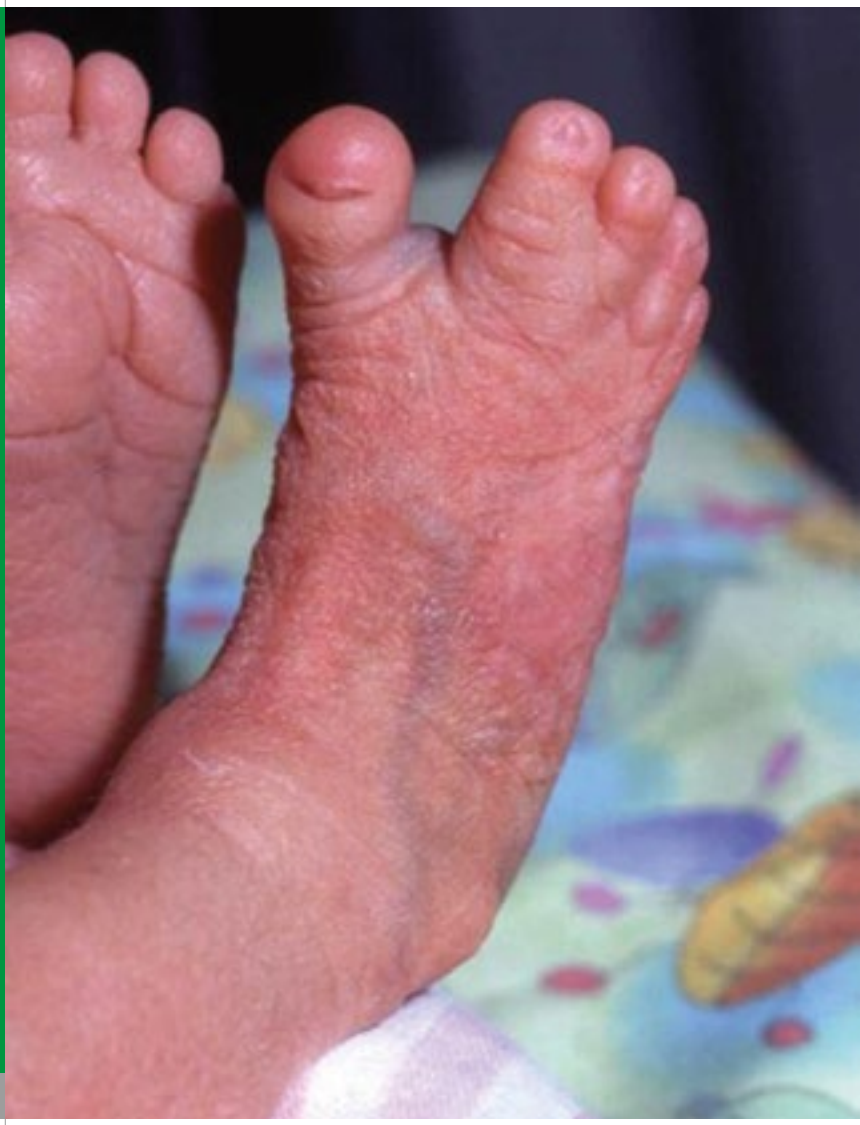

Fig. 7

Syndactylies and distal hypoplasia of toes.

Yunis-Varon syndrome was first described in 1980 by Emilio Yunis and Humberto Varon in five children from three families. Because of parental consanguinity and affection of both sexes, the authors suggested an autosomal recessive inheritence. In the meantime, 13 cases have been published with, in part, divergent clinical features.

The syndrome is characterised by pre- and postnatal growth restriction, combined with severe mental retardation. Dysmorphic features include microcephaly, sparse scalp hair (including eyelashes and eyebrows), wide fontanels with diastasis of sutures, thin lips, a short philtrum, labiogingival retraction, anteverted nostrils, abnormally modelled low-set ears and redundant posterior neck skin. Skeletal manifestations include absent or hypoplastic thumbs and halluces, short and pointed fingers and toes with nail hypo-/aplasia, distal aphalangia and syndactylies, as well as micro-/retrognathia, hypo-/aplasia of the clavicles, abnormal scapulae, missing sternal ossification, pelvic dysplasia and hip luxation.

In some patients, additional abnormalities have been described such as CNS malformations (aplasia of corpus callosum, cerebellar hypoplasia, hydrocephalus), cardiac malformations (Fallot tetralogy, VSD). Absent nipples, cataracts and atrophy of the left liver lobe have also been reported.

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