Severe intrauterine growth retardation, blepharophimosis, and cylindrical nose with midline groove: a new syndrome?

C E M de Die-Smulders, R P Droog, M van Dijk, J P Fryns

Abstract

A malformed female infant is described. In addition to cardiac, renal, and skeletal (rib) anomalies, severe intrauterine growth retardation and distinct facial dysmorphism were present. The question is raised whether this child represents a new syndrome.

(*J Med Genet* 1993;30:525)

Case report

The female patient is the third child of healthy non-consanguineous parents. The two older sisters are normal. Severe growth retardation became progressively evident from the beginning of the second trimester of pregnancy. Amniocentesis showed a normal female karyotype. Ultrasound examination was performed at 35 weeks and showed, in addition to the growth retardation (growth equivalent to 27 weeks), a complicated cardiopathy (complete AV channel), facial anomalies (hypertelorism, broad nose, micrognathia), and flexed position of both hands. Labour was induced at 39 6/7 weeks. The girl died immediately after birth because of severe respiratory distress. Birth weight, length, and head circumference were

1160 g (3rd centile = 2400 g), 41 cm (3rd centile = 46 cm), and 25.7 cm (3rd centile = 31.5 cm) respectively. The craniofacial appearance was peculiar with a small, triangular face, a high and narrow forehead with low hair implantation, a broad nasal bridge, short, narrow palpebral fissures, bilateral epicanthus, cylindrical, beaked nose with a median groove, a deep philtrum, thin lips, a small mouth, and micrognathia (figs 1 and 2). Both ears were low set and the left ear was poorly lobulated and everted (cup ear). The neck was rather short with lateral webbing. The thorax was narrow in its upper part. The genitalia were normal female. Both hands were flexed to 40° and radially deviated. Foot length (6 cm) was proportionate to the other body measurements. Necropsy showed the following internal malformations: complete AV channel, coarctation of the aorta, hypoplastic right external iliac artery, aplasia of the right umbilical artery, pelvic position of the right kidney, dilated left ureter with dysplasia of the ureterocystic junction. Hypoplasia of the 2nd to 5th ribs on the left and the 1st and 3rd rib on the right was seen on x ray.

Discussion

This malformed female infant had a unique combination of clinical signs and symptoms comprising severe intrauterine growth retardation, unusual facial appearance, and internal malformations. The facial stigmata were distinct with a narrow forehead, flat orbital ridges, narrow palpebral fissures, and bilateral epicanthus. The nose was particularly abnormal; it was beaked with a broad nasal bridge and the top was broad with a median groove. Because of the facial features in this child the diagnosis of Seckel syndrome and other microcephalic-primordial dwarfism syndromes is not very likely. Among the different syndromes with marked intrauterine growth retardation as a cardinal symptom Dubowitz syndrome was considered.^{1,2} However, the growth retardation in the proband of this report is much more severe, the facial anomalies are not typical, and the cardiac abnormalities are more severe than described in Dubowitz syndrome. We suggest that the present patient could be the first instance of a hitherto undelineated severe intrauterine growth retardation syndrome.

1 Dubowitz V. Familial low birth weight dwarfism with an unusual face and a skin eruption. *J Med Genet* 1965;2:12-17.

2 Winter RM. Dubowitz syndrome. *J Med Genet* 1986;23:11-13.

Department of Clinical Genetics, Academic Hospital Maastricht, PO Box 1475, 6201 BL Maastricht, The Netherlands.
C E M de Die-Smulders

Department of Paediatrics, Elkerliek Hospital, Helmond, The Netherlands.
M van Dijk

Department of Pathology, Elkerliek Hospital, Helmond, The Netherlands.
R P Droog

Department of Human Genetics, Academic Hospital, Gasthuisberg, Leuven, Belgium.
J P Fryns

Correspondence to Dr de Die-Smulders.

Received 12 May 1992.
Revised version accepted 15 October 1992.

Figure 2 The nose with a broad tip and median groove.

Figure 1 The distinct facial dysmorphism.