Dubowitz syndrome

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This syndrome was first described 20 years ago by Professor Victor Dubowitz in this Journal.1 Since then about 30 possible cases have been reported, including a follow-up of one of Dubowitz's cases.2

The main features of the condition are intrauterine growth retardation, primordial short stature, eczematous skin eruption, and a characteristic facies. The facial abnormalities make up a recognizable gestalt, and are described below and illustrated in the photographs. Particularly good reviews of the condition are to be found in Grosse et al.,2 Opitz et al.,3 and Wilroy et al.4

Clinical features

GROWTH
At birth the average weight is 2.3 kg, the average length is 45 cm, and the average head circumference is 30 cm. Postnatal growth deficiency is usual, but not severe. Delayed bone age has been reported in approximately 50% of cases.

CRANIOFACIAL
The main features are microcephaly (100%), sparse hair (70%), sloping forehead (80%), telecanthus (60%), ptosis/blepharophimosis (often asymmetrical) (65%), epicanthic folds (50%), broad nose (55%), palate anomalies (high/narrow/cleft/submucous cleft) (50%), micrognathia (80%), high pitched/hoarse voice (55%), and prominent or mildly dysplastic ears (75%).

The facial appearance changes with age, but micrognathia (with a pointed chin) appears constant.
at all ages. However, the contour of the face elongates with age and the nasal bridge becomes more prominent; in older patients it is high and almost continuous with the forehead. The base of the nose is broad. These changes are best illustrated in the paper by Grosse et al. The supraorbital ridges are hypoplastic with arched eyebrows, sparse laterally. Facial eczema may be present.

**BEHAVIOUR/DEVELOPMENT**

Prominent features are poor feeding (55%), vomiting and diarrhoea in early infancy (40%). hyperac-
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tivity (35%), and mild to moderate mental retardation (50%). Parrish and Wilroy\(^5\) found one case out of 10 to have severe mental retardation. Short attention span and impulsivity were also noted.

OTHER FEATURES
Those reported include eczema in infancy (50%), hypospadias/cryptorchidism (40% of males), syndactyly of the second and third toes (40%), aplastic anaemia (two cases\(^6\)), immunodeficiency and malignant lymphoma/neuroblastoma (two cases\(^7\)), acute lymphatic leukaemia (one case\(^8\)), and aplastic anaemia (two cases\(^6\)).

Inheritance
The existence of an equal sex ratio with at least six sets of affected sibs with normal parents, and at least one case of parental consanguinity,\(^3\) makes autosomal recessive inheritance highly likely.

Differential diagnosis
Differential diagnosis includes other causes of intrauterine growth retardation, especially the fetal alcohol syndrome and Bloom syndrome.

Note added in proof
Moller and Gorlin\(^9\) have published a follow up of the two sibs originally described by Grosse et al,\(^2\) together with a further review of the condition.

References

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