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Syndrome of the month

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Wiedemann–Rautenstrauch syndrome

Helga V Toriello

In 1977 Rautenstrauch and Snigula[1] reported on two sisters with a previously unreported, progeria-like syndrome. In 1979 Wiedemann[2] described two unrelated males with the same condition, in 1981 Devos *et al*[3] described another affected child whose parents were double first cousins, and in 1988 Rudin *et al*[4] reported on a single affected child, bringing the total number of reported patients to six. Martin *et al*[5] described neuropathological studies on the child described by Devos *et al*[3] and suggested that the Wiedemann-Rautenstrauch syndrome is a form of sudanophilic leucodystrophy.

Clinical features

All affected children have had intrauterine growth retardation with subsequent failure to thrive and short stature. At birth and during infancy children have a progeroid appearance consisting of apparent macrocephaly (although occipitofrontal circumference is usually within normal limits for age), sparse hair, prominent scalp veins, entropion, greatly widened anterior fontanelles, malar hypoplasia, and generalised lipoatrophy (figs 1 and 2). Two to four natal teeth have been present in all affected children; these teeth are eventually lost and subsequent dentition is delayed. Over time, the nose assumes a beaked appearance and caudal fat accumulation occurs (fig 3). Mental retardation is usually present to some degree, ranging from mild to severe impairment. Hypotonia, truncal ataxia, and intention tremor may also be present. Longevity is unknown, although the oldest reported child died at age 5½ of bronchopneumonia.

Radiographical studies have shown partial non-ossification of the atlas, hypoplastic iliac bones with a trident configuration of the acetabula, and irregular ossification of the ilia. These findings become normal by 1 year of age.[4]

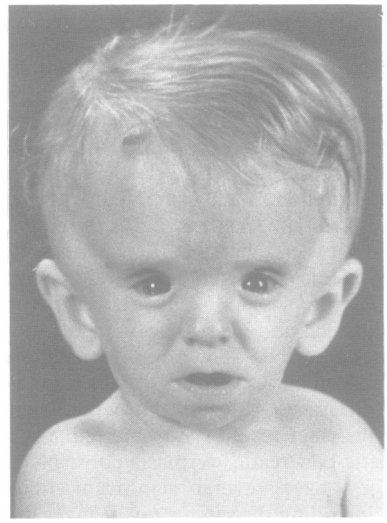

Figure 1 Patient 2 reported by Wiedemann[2] aged 8 months. Note apparent macrocephaly, prominent scalp veins, entropion, and malar hypoplasia.

Differential diagnosis

Differential diagnosis includes other sudanophilic leucodystrophies, such as congenital Pelizaeus-Merzbacher disease, Sjögren-Larsson syndrome, Cockayne syndrome, sudanophilic leucodystrophy with microcephaly and pachygyria, as well as progeroid or lipoatrophy syndromes such as Hutchinson-Guilford, Berardinelli-Seip, DeBarsy, and Hallerman-Streiff syndromes. However, the striking phenotype and presence of natal teeth help distinguish this condition from others.

Natural history and treatment

These children often suffer from feeding difficulties

Genetics Services, 21 Michigan Street, Suite 350, Grand Rapids, Michigan 49503, USA.
H V Toriello

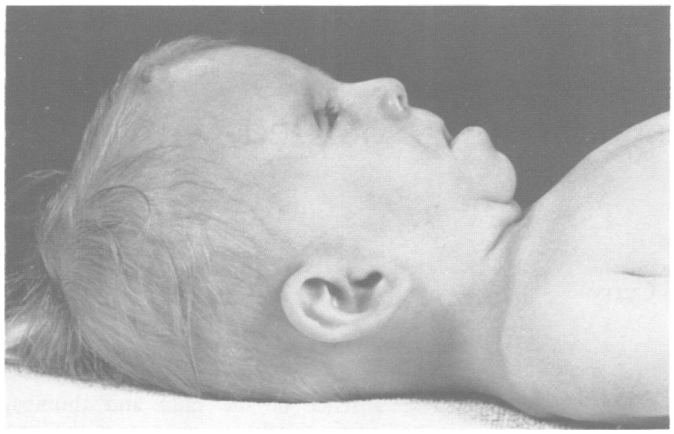

Figure 2 Same patient, side view.

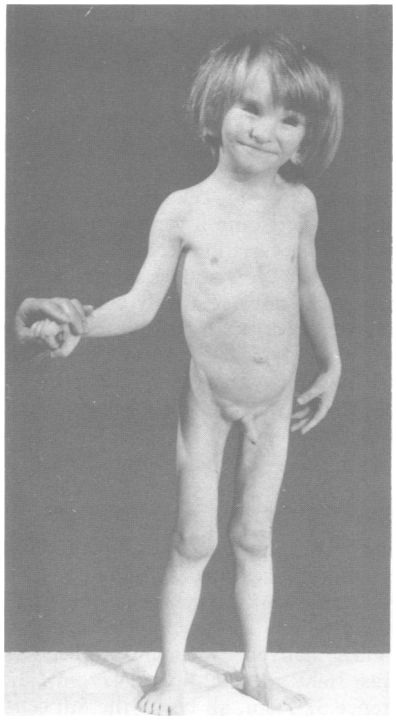

Figure 3 Same patient, aged 3 years. Note lipoatrophy.

and respiratory tract infections. Short stature is always ultimately present; the patients described by Wiedemann[2] were markedly growth deficient, whereas those described by Rautenstrauch and Snigula[1] had normal growth at 8 months of age. However, in a follow up report of their patient, Snigula and Rautenstrauch[6] noted that by the age of 4 years, one of their patients was markedly growth deficient.

Children with this syndrome may also be more prone to intracranial haemorrhage, particularly after head trauma.

Inheritance

This condition is most probably inherited as an autosomal recessive trait, in that Rautenstrauch and Snigula[1] described affected sibs and Devos *et al*[3] described a child whose parents were consanguineous.

Developmental basis and aetiology

Martin *et al*[5] described neuropathological changes consistent with sudanophilic leucodystrophy. The white matter showed a sudanophilic demyelinisation which had a faint tigroid pattern. Polymicrogyric patches were found in the cortex. However, the peripheral nervous system was intact.

Professor Wiedemann kindly made helpful suggestions for the manuscript and supplied the illustrations.

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2 Wiedemann HR. An unidentified neonatal progeroid syndrome: follow-up report. *Eur J Pediatr* 1979;**130**:65–70.
3 Devos EA, Leroy JG, Fryns JP, Van den Berghe H. The Wiedemann-Rautenstrauch or neonatal progeroid syndrome. Report of a patient with consanguineous parents. *Eur J Pediatr* 1981;**136**:245–8.
4 Rudin C, Thommen L, Fliegel C, Steinmann B, Buhler U. The neonatal pseudo-hydrocephalic progeroid syndrome (Wiedemann-Rautenstrauch). *Eur J Pediatr* 1988;**147**:433–8.
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6 Snigula F, Rautenstrauch T. A new neonatal progeroid syndrome. *Eur J Pediatr* 1981;**136**:325.