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NEONATAL PROGEROID SYNDROME: REPORT OF A JAPANESE INFANT

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Summary A Japanese male infant with the neonatal progeroid syndrome is described, bringing to six the number of patients reported with the syndrome. He had clinical features typical of the syndrome, and in addition, several abnormalities not described previously. The latter included blepharophimosis, downward slanting of the palpebral fissures, long eyelashes, narrow palate, generalized articular contractures, camptodactyly of the 2nd through 4th fingers, and the right second toe overriding the great toe.

INTRODUCTION

The neonatal progeroid or Wiedemann-Rautenstrauch syndrome is a syndrome delineated by Wiedemann (1979), based on the observation of two sisters reported by Rautenstrauch *et al.* (1977), and of two unrelated patients of his own. The syndrome is characterized by progeroid face and generalized diminution of the subcutaneous fat, both apparent from birth, and developmental retardation. Autosomal recessive inheritance has been suggested based on its occurrence in sisters and in a girl born to consanguineous parents (Devos *et al.*, 1981). We will here describe a Japanese male infant with clinical features typical of the syndrome, and with several additional abnormalities not described previously.

CASE REPORT

F.H. (0223-25-2), a male, was born after an uneventful pregnancy of 39 weeks to a 33-year-old, gravida V, para II mother and a 32-year-old-father, both healthy and unrelated. His two elder brothers, 2 and 7 years old, respectively, were normal. The mother had experienced a spontaneous abortion and an induced abortion.

There was no family history of malformations. At birth, the patient weighed 2,300 g (−2.6 SD), measured 48.5 cm (−0.72 SD) and his head circumference was

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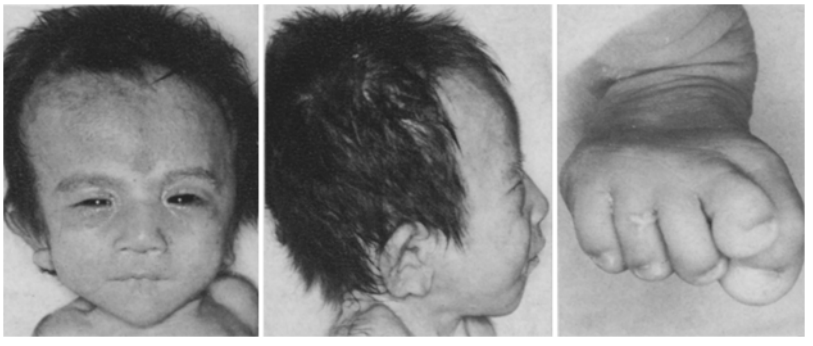

Fig. 1. The patient at age one month. A large cranium, dilated scalp veins, hypoplasia of the facial bones, thin eyebrows, downward slanting of the palpebral fissures, blepharophimosis, a high nasal bridge, micrognathia (left and middle), and the right second toe overriding the great toe (right).

30 cm (−2.6 SD). His umbilical cord was long at 104 cm. A progeroid facial appearance was noted at birth together with generalized absence of the subcutaneous fat. Other features noted were hirsutism (especially on the shoulders and back, and with tufts of the hair under the temples), a relatively large cranium with small facial bones, prominent scalp veins, thin eye brows, long eyelashes, downward slanting of the palpebral fissures, blepharophimosis, a high nasal bridge, a narrow palate, micrognathia, a large penis, generalized articular contractures, camptodactyly of the index through ring fingers and the right second toe overriding the great toe (Fig. 1).

Soon after birth, he showed signs of congestive heart failure with hepatomegaly. Echocardiography revealed patent ductus arteriosus (PDA). Digitalization was started. The PDA closed on 9 days of life after 2 days' indomethacin administration, but secondary endocardial fibroelastosis developed with left ventricular dysfunction. Computed tomographic examination of the brain at age one month revealed dilatation of the ventricles. G-banded chromosomes in cultured PB lymphocytes were normal. He suffered from feeding difficulties and frequent vomiting. His growth and psychomotor development were severely retarded. He could not control his head or roll over at age 18 months. His length was 63 cm (−6.2 SD), weight 4,810 g (−5.4 SD) and head circumference 40.6 cm (−4.6 SD). His external appearance was nearly unchanged (Fig. 2). The hair was sparse, the large fontanel measured 2.6 × 3.0 cm, and the subcutaneous veins, especially the scalp veins, were prominent because of generalized diminution of the subcutaneous fat. He died at age 19 months of suffocation from vomited milk. Autopsy was not granted.

DISCUSSION

The neonatal progeroid syndrome is apparently infrequent. Only five patients

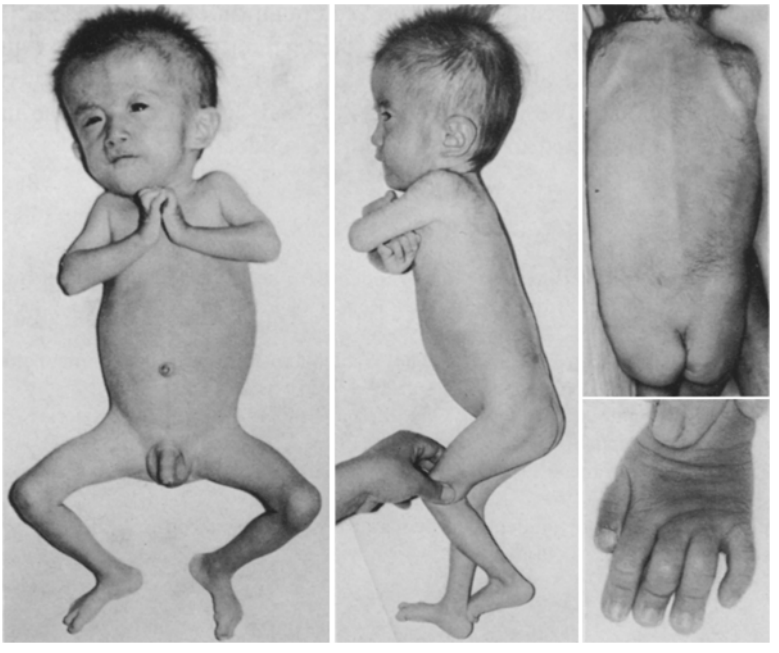

Fig. 2. The patient at age 12 months, with essentially unchanged clinical features. Generalized diminution of the subcutaneous fat, a sparse scalp hair, hirsutism, a large penis (left and right), narrow hips, absence of caudal fat accumulation (right top), and camptodactyly of the index through ring fingers (right bottom).

have been reported, all from Europe. Rautenstrauch *et al.* (1977) of Munich described two sisters with the disease, although they considered it as classical progeria. The same group later published a follow-up account of the younger, surviving sister (Snigula and Rautenstrauch, 1981). Wiedemann (1979) of Kiel reported on two sporadic male infants and coined the term "neonatal progeroid syndrome." Devos *et al.* (1981) of Belgium described a 4-year-old girl born to parents who were double first cousins. The patient died at 5 1/2 years of age. Her neuropathological data were presented in a separate paper, which indicated an extensive demyelination of the central nervous system (Martin and Ceuterick, 1984).

The clinical features common to these five patients and the patient we described included progeroid face apparent from birth, generalized diminution of the subcutaneous fat, growth and developmental delay, hydrocephaloid skull, persistent fontanel, prominent scalp veins, sparse scalp hair, hypoplastic facial bones, high nasal bridge and large penis. Clinical features present in our patient but not described previously included antimongoloid slanting of the palpebral fissures, blepharophimosis, long eyelashes, camptodactyly of the 2nd through 4th fingers, the right second toe overriding the great toe, generalized congenital articular contractures, hirsutism and PDA. Features found in the five previous patients but not present in

our patient were teeth present at birth and accumulation of subcutaneous fat patches in the dorsocaudal region. In our patient, the teeth erupted at age 5 months.

It is noteworthy that all five previous patients reported were of either German or Belgian extraction. The occurrence of the syndrome in a Japanese infant indicates that it is not restricted to individuals in these racial groups.

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