

therapy for atherosclerosis and coronary artery disease in these patients becomes more effective, more progeria patients may be identified with malignancy.

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The Aase syndrome in a female infant

SUMMARY This report describes a 2-month-old female with the Aase syndrome, bringing to 8 the total number of cases of this disorder. Features include triphalangeal thumbs and congenital hypoplastic anaemia. The occurrence of this disorder in sibs born to unaffected parents and in both sexes makes autosomal recessive inheritance the most likely aetiology.

This report describes a female infant with triphalangeal thumbs and congenital erythroid hypoplasia. Seven similar cases of this disorder, referred to as the Aase syndrome, have been described (Harvey, 1966; Aase and Smith, 1969; Murphy and Lubin, 1972; Jones and Thompson, 1973; Terheggen, 1974; van Weel-Sipman *et al.*, 1977).

Case report

The patient was a 2-month-old Mexican female. She was born to a 20-year-old, gravida 1 woman after an uncomplicated 40 week gestation; birthweight was 3.5 kg. Length and head circumference were 51 cm and 35.5 cm, respectively. Paleness and progressive lethargy were noted at 6 weeks of age. She was referred at 2 months of age for evaluation of severe anaemia. Weight was 4.3 kg (25th centile for age), length was 54 cm (25th centile), and head circumference was 38 cm (50th centile). Positive physical findings included a grade 2/6 systolic ejection murmur at the lower left sternal border, and striking hand abnormalities consisting of digitalised thumbs and hypoplastic thenar eminences (Fig. 1). Dermatoglyphs were normal except for a horizontal pattern over the thenar areas.

Haematological evaluation showed: haemoglobin 4.3 g/dl, haematocrit 14%, reticulocyte count 0.8%, white blood cell count $5.8 \times 10^9/l$ with 33% neutrophils, 7% bands, 39% lymphocytes, 16% mononuclear cells, and 5% eosinophils. Platelet count was $600 \times 10^9/l$. Bone marrow showed a pure red cell aplasia with a myeloid to erythroid ratio of 75 to 1 and normal numbers of megakaryocytes. Studies of cultured bone marrow showed a normal 46,XX karyotype, with no evidence of chromosomal breakage such as has been demonstrated in the Fanconi pancytopenia syndrome. Significant radiographic abnormalities included a triphalangeal right thumb, hypoplasia of the left thumb (Fig. 2), and a single bifid thoracic vertebra. Cardiac evaluation, including chest

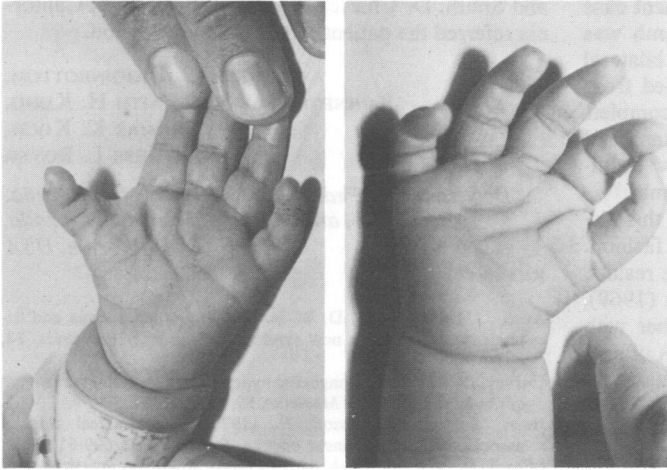


Fig. 1 Left and right hands. Note thumb deformities, thenar hypoplasia, and altered palmar crease patterns.

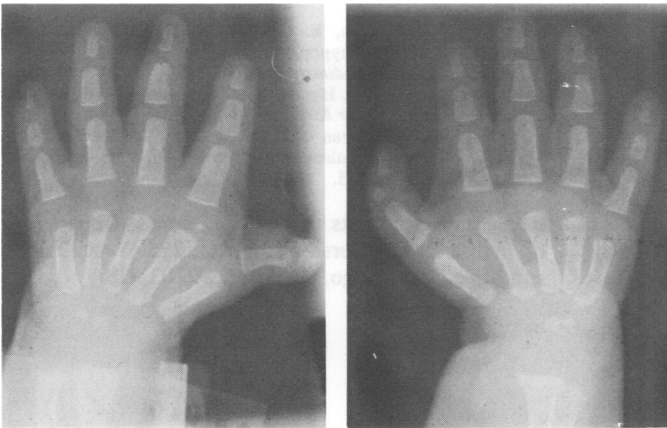


Fig. 2 X-rays of left and right hands. Note triphalangeal thumb on the right, and hypoplasia of the distal and proximal phalanges of the thumb on the left.

x-ray, electrocardiogram, and echocardiogram, showed no significant abnormality. Electrolytes and renal and liver function tests were normal. The child was transfused with packed red blood cells and started on 2 mg/kg prednisone per day.

The parents of the patient were normal and denied consanguinity. However, they are both from the same small town in Mexico.

Discussion

The present patient brings to 8 the number of cases described with the Aase syndrome. All children with this condition have developed severe anaemia in the first year of life. The age of presentation, the lack of

chromosomal breakage on cultured leucocytes, and the relative sparing of the granulocytic and platelet precursors distinguish this disorder from the Fanconi pancytopenia syndrome. Moreover, the radial defects in the 2 conditions are different. The Aase syndrome is characterised by triphalangeal thumbs with minimal to no involvement of the radius. In the Fanconi pancytopenia syndrome an array of abnormalities from hypoplasia to aplasia of the thumb, radius, carpal, and metacarpal bones has been described. Thumb duplications also occur but triphalangeal thumbs have not been seen (Minagi and Steinbach, 1966).

That there is a spectrum to both the thumb abnormalities and the degree of erythroid hypoplasia is apparent from a comparison of the existing cases. All 7 of the previously described patients have had tri-

phalangeal thumbs on both hands. In the present case radiographic evidence of a triphalangeal thumb was shown only on the right hand, in spite of bilateral deformities. Bone marrow findings have varied from the present patient, who exhibited almost complete lack of erythroid precursors, to case 1 of Aase and Smith (1969), who had a normal bone marrow examination despite the presence of significant anaemia.

The response to prednisone in patients with this disorder has not been evaluated in a controlled fashion. However, there have been some encouraging results. Of the 2 patients reported by Aase and Smith (1969), one was treated with prednisone and the other with blood transfusion alone. Now 13 and 10 years of age, neither child needs steroids or blood transfusion to maintain a haematocrit in the high 20s. These children, however, did not exhibit the severe degree of erythroid hypoplasia seen in the present patient. Severe red cell aplasia was shown in the case reported by Jones and Thompson (1973) and the case of van Weel-Sipman *et al.* (1977). The anaemia in the former patient was prednisone responsive over the 8 years of follow-up described in the report. The anaemia in the latter case failed to respond significantly to steroids, and over 9 months of follow-up continued to require repeated blood transfusions.

Of the 8 patients described, 5 have been male and 3 female, and 2 have been sibs. Autosomal recessive inheritance is the most likely aetiology.

Dr Robert Hickman, Seattle, Washington generously provided follow-up information on the 2 cases of Aase

and Smith. Dr Charles Freedman, San Diego, California referred the patient reported for evaluation.

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