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CASE IMAGES

Infant with a big head and 'crossed' polysyndactyly

Question

A 10-week-old boy was referred to our hospital for polysyndactyly of all four limbs and frontal bossing. He was born at term by emergency caesarean section for pre-eclampsia and breech presentation with Apgar scores of 9 and 10 at 1 and 5 min. Family history was unremarkable and prenatal obstetric ultrasounds were regular. Neonatal cerebral ultrasound, otoacoustic emissions, echocardiography and abdominal ultrasound were all normal. No prenatal or post-natal genetic analysis was performed.

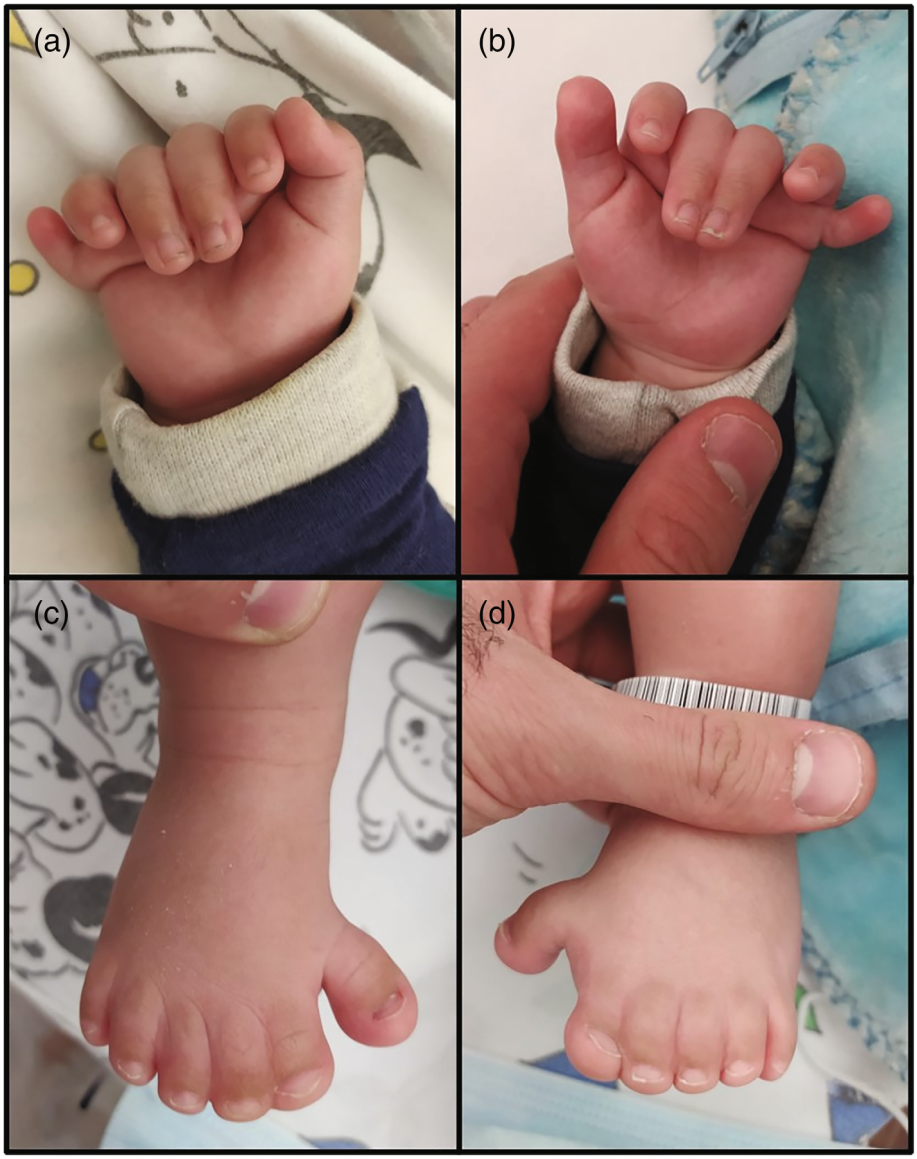

Fig. 1 'Crossed polysyndactyly': post-axial polydactyly with syndactyly of third and fourth fingers of both hands (a, b) and pre-axial polydactyly with syndactyly of all the non-supernumerary toes of both feet (c, d).

Fig. 2 Frontal bossing with head circumference just above the upper limit of normal.

On physical examination, post-axial polydactyly and syndactyly of the third and the fourth fingers of both hands (Fig. 1a,b) were noted, as well as pre-axial polydactyly and syndactyly of all the non-supernumerary toes of both feet (Fig. 1c,d). This distinctive combination of hands and feet abnormalities constitutes the so-called 'crossed polysyndactyly'. The infant also had frontal bossing, and his head circumference measured 42 cm (slightly above 97° centile, according to the WHO charts) with an open anterior fontanelle (Fig. 2). Neurological development appeared appropriate for age, and the growth rate was average.

What is the likely diagnosis? (Answer on page 773)

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Pre-school girl with bluish purple fingers

Question

A previously well 5-year-old girl, presented to a paediatric clinic in central west New South Wales with swollen, sore, red-blue-purple fingers, hands and toes of 4 months duration (Fig. 1). Onset of symptoms coincided with beginning of winter and, although waxing and waning of skin discolouration was noticed, swelling of fingers and toes persisted.

She was appropriately grown, systemically well and there was no family history of autoimmune or connective tissue disease. What is the likely diagnosis (Answer on page 774)?

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