

IMAGES IN DERMATOLOGY

Aplasia Cutis Congenita Associated with Trisomy 13

Aplasia cutis congénita asociada a trisomía 13

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A 38-week gestation male was diagnosed with trisomy 13 by amniocentesis and was found to have hydronephrosis by prenatal ultrasound.

At birth the patient presented APGAR scores of 6-8, weak breathing, a grade two holosystolic murmur located on the left parasternal line and a well-circumscribed absence of skin, about 5 cm of diameter, on the midline vertex (Fig. 1).

His 35-year-old mother denied a family history of aplasia cutis. The findings from the remainder of the examination were normal. General blood tests were normal, chest X-ray showed cardiomegaly.

A dermatology consultation was performed and the patient was diagnosed as having aplasia cutis congenita.

Aplasia cutis congenita can be classified into nine different groups. The ninth group is associated with malformation syndromes including trisomy 13 (Patau syndrome). Because of the poor prognosis of Patau syndrome the patient was managed conservatively, with no specific investigation or



Figure 1

treatment performed on the patient. The patient died after 5 days, most likely due to renal or cardiac disease.

Central nervous system or cranial involvement must be ruled out in cases of aplasia cutis that have the presence of hair collar sign, midline vertex location, size greater than 5 cm, vascular stains and nodules.

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