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Clinical Image

CLINICAL IMAGE

Congenital primary cutis verticis gyrata

Veronica Mugarab-Samedi[1], Salam Al Jubouri[1] and Alixe Howlett[1,2]

[1]Division of Neonatology, University of Calgary, Calgary, Canada, [2]Division of Neonatology, Alberta Children's Hospital, Calgary, Canada

*Correspondence address. 780, 1403 29th Street NW, Calgary, AB T2N 2T9, Canada. Tel: 403 3971762; Fax: 1-403-955-7442; E-mail: vmsamedi@gmail.com

Congenital cutis verticis gyrata (CCVG) affects 1 in 100 000 males and 0.026 in 100 000 females. Characterized by thick skin folds and grooves on the scalp resembling the outer surface of the brain (1), CCVG can present in primary and secondary forms (2). The inheritance of this condition remains controversial (3). While primary CCVG has been associated with chromosomal abnormalities as Noonan and Turner syndromes, there are no many reports of CCVG in a patient with Trisomy 21 (4). Timely recognition of CCVG is important as this condition is linked to serious neurological and ophthalmological anomalies.

Our patient was born to a healthy 36-year-old primigravida Asian mother. The pregnancy was uneventful until 29 weeks of gestational age when she went to spontaneous labour and delivered by unassisted vaginal delivery a 1150-gm male infant. On initial assessment, among typical features of trisomy 21 (flat occiput, flattened facial appearance, small brachycephalic head and epicanthal folds), deep cerebriform folds and wrinkles over the entire scalp more prominent in the parietal-occipital region were noted (Fig. 1). On palpation the area was spongy, with no erythema, infiltration or skin defects. Cranial and abdominal sonography, ophthalmological assessment were normal. Genetics and dermatology were consulted, and the condition was diagnosed as CCVG. Diagnosis of trisomy 21 was confirmed by microarray. The infant had an uneventful hospital course and was discharged to home after 6 weeks. No further investigations or interventions for this condition were required. At his 12-month assessment there has been no progression of the condition noted.

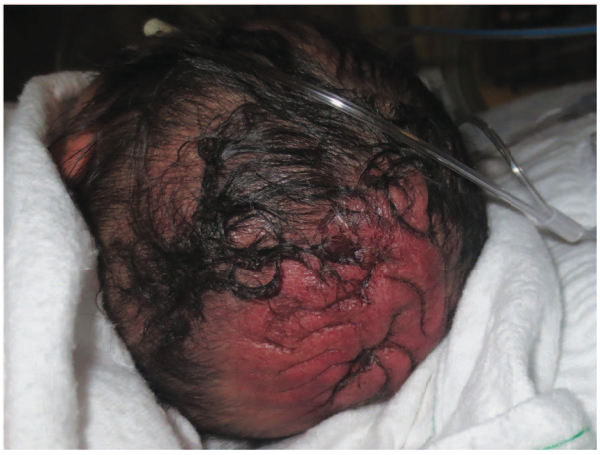

Figure 1: Patient at the first week of life showing the asymmetrical skin folds extending from the occiput to the scalp.

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CONFLICT OF INTEREST

None declared.

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ETHICAL APPROVAL

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CONSENT

Confirmation of written consent from patient/guardian: parental consent provided.

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