Temporal skin folds in a female infant with an unbalanced translocation with breakpoints Xq22.1 and 6p22.3: a new association?

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DESCRIPTION

A female infant was born to healthy unrelated parents with no relevant family history. She had normal antenatal scans and was born by spontaneous vaginal delivery at 33 weeks' postmenstrual age. She was admitted to the neonatal unit for prematurity and respiratory distress. She was noted to have marked bilateral and dysplastic skin folds on her temples (figures 1 and 2) but no other dysmorphic features.

Array comparative genomic hybridisation and subsequent fluorescence in situ hybridisation detected an unbalanced X autosome translocation resulting in 53 Mb deletion at Xq22.1q28 and 17.5 Mb duplication at 6p22.3p22.3. Both parents had normal karyotypes indicating that the translocation had arisen de novo. Thyroid function tests, structural/functional echocardiography, cranial and renal ultrasound scans and ophthalmological examination were normal. Hearing test was also within normal results.

At the age of 14.5 months neurodevelopmental assessment (Bayley III) was performed.¹ After adjustment for prematurity, the performance was within the expectations for age: Cognitive Composite Score 85 (percentile rank 16), Language Composite Score 103 (percentile rank 58) and Motor Composite Score 103 (percentile rank 58). Neurological examination was normal and there were no clinical concerns.

The translocation did not have exact precedent in the medical literature. As with other X autosome translocations in females, its phenotypic impact may be ameliorated by non-random X inactivation.

Figure 1 Left temporal skin folds.

Figure 2 Right temporal skin folds.

A small number of individuals have been reported with similar focal facial dermal dysplasias such as Setleis syndrome.² The case does not establish a causal relationship, but reports a possible new association between this translocation and temporal skin folds.

Learning points

- ▶ We report a child with bilateral temporal skin folds and with an unbalanced translocation with breakpoints at Xq22.1 and 6p22.3, a possible novel association.
- ▶ Unbalanced translocation with breakpoints at Xq22.1 and 6p22.3 is not associated with impaired early neurodevelopmental outcome.
- ▶ Array comparative genomic hybridisation is the first line investigation for the detection of unbalanced chromosome translocations.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCES

- 1 Bayley N. *Bayley scales of infant and toddler development-II*. San Antonio, Texas: The Psychological Corporation, 2006.
- 2 Garcia-Minaur S, Linares A. Setleis ("bitemporal forceps marks") syndrome. *Clin Dysmorphol* 1995;4:173–5.

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