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## **Spectral Karyotyping (SKY) permits the Characterisation of a de Novo unbalanced Translocation 46, XY, der(14)t(12;14)**

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**KEYWORDS** Spectral karyotyping; SKY; unbalanced translocation 12/14 de novo; dup 12q24; euchromatin-heterochromatin rearrangement

**ABSTRACT** A boy with a duplication 12q24 to qter shows the characteristic phenotype of this chromosome abnormality. The aberration could be analysed as an unbalanced translocation 12/14 de novo by combining different banding techniques with FISH and SKY. Heterologous euchromatin translocations on the heterochromatic short arm regions of the acrocentrics are an extremely rare type of rearrangement.

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