Appendix S1.

MATERIALS AND METHODS

Multiplex ligation-dependent probe amplification (MLPA)

MLPA analysis using a SALSA P018-E1 kit (MRC Holland, Amsterdam, The Netherlands) for the detection of deletions or duplications in each exon of the *SHOX* gene and the 250-kb downstream regulatory domain was performed according to the manufacturer's protocol (http://www.mrc-holland.com/WebForms/WebFormMain.aspx). Of note, additional probes located in and outside of the pseudo-autosomal region 1 (PAR1) as well as in the *NLGN4X* (Xp22.32) or *KAL1* (xp22.31) genes were designed. The SALSA P018-E1 kit is based on the method described by Schouten et al. (1).

Real-time quantitative PCR

Using the LightCycler® 480 Real-Time PCR System (ROCHE, Mannheim, Germany), DNA extraction was performed according to the manufacturer's protocol for whole blood samples. The *STS* and *VCX3A* gene deletion patterns were analysed by PCR. The conditions and primers used to analyse the *VCX3A* and *STS* genes are described elsewhere (2). Information about the primers used is available from the authors.