

**Figure S2: Chromosomal microarray analysis (CMA) of subject III:2**

- A. (Upper) CMA profile of chromosome 8 (left) and magnified view of plot array showing the 8q24.3 deletion of ~124 kb (right).  
 (Bottom) The deleted region aligned with UCSC map (hg19) in red and genes are indicated. Interpretation of rearrangement according to ISCN 2016: arr[GRCh37] 8q24.3(140811012x2,140821366\_140945585x1,140964076x2).
- B. (Upper) CMA profile of chromosome X (left) and magnified view of plot array showing the Xp22.2 deletion of ~58.9 kb (right).  
 (Bottom) The deleted region aligned with UCSC map (hg19) in red and genes are indicated. Interpretation of rearrangement according to ISCN 2016: arr[GRCh37] Xp22.2(13505438x1,13551079\_13610003x0,13629731x1).

