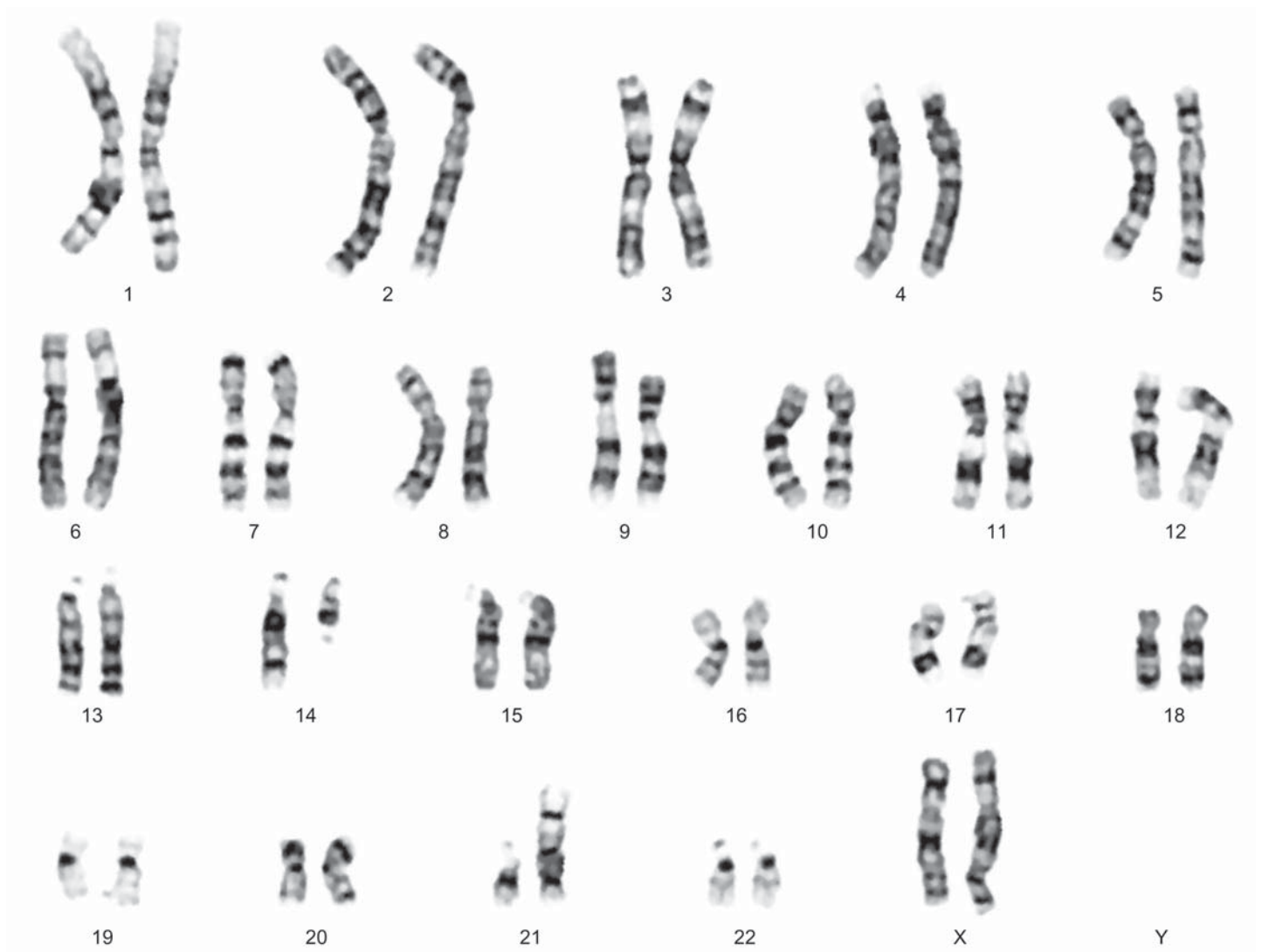


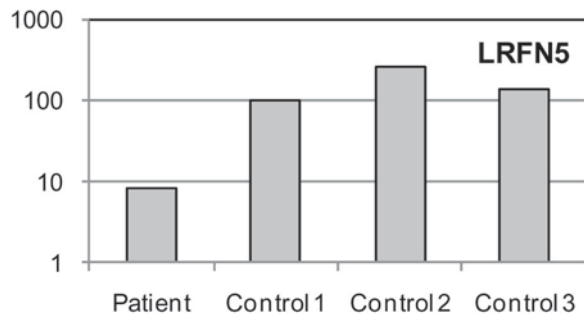
Supplementary Fig. 1

Pictures of the patient taken at ages 9 months and 6 years.



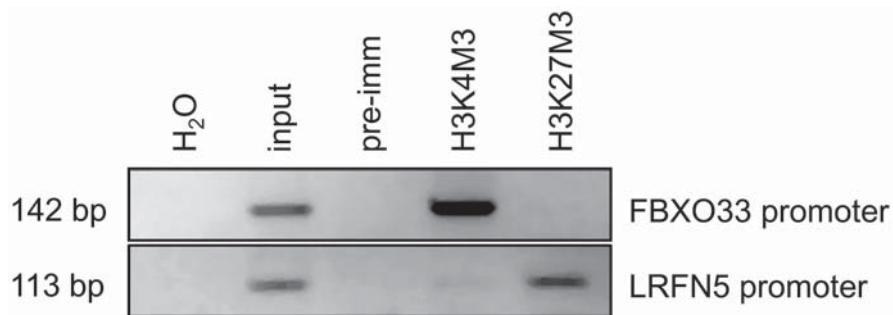
Supplementary Fig. 2

G-banded chromosomes showing the balanced translocation between chromosomes 14 and 21, karyotype 46,XX,t(14;21)(q21.1;p11.2).



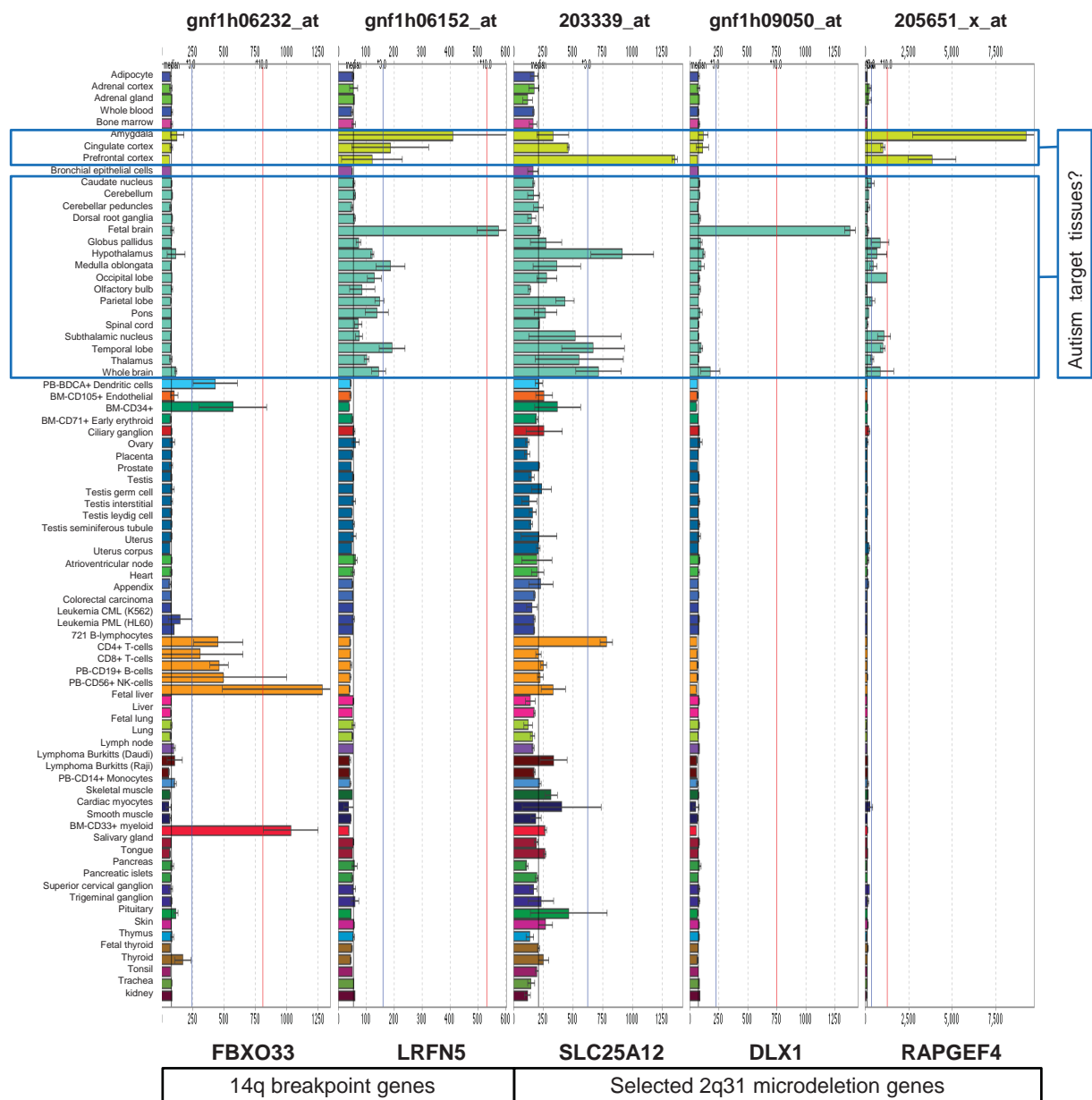
Supplementary Fig. 3

Expression analysis of LRFN5 in the patient and three control fibroblasts. LRFN5 expression was readily detected in the control fibroblasts, but was reduced tenfold in the patient fibroblasts. Bar graphs show the level of LRFN5 expression determined by real-time RT-PCR, the scale is logarithmic.



Supplementary Fig. 4

Semi-quantitative PCR for the FBXO33 and LRFN5 promoters on amplified patient fibroblast ChIP material. These data show that the FBXO33 promoter is positive for H3K4M3, but not for H3K27M3. In contrast, the LRFN5 promoter is positive for H3K27M3 and very weakly positive for H3K4M3. These results are in support of the ChIP-on-ChIP data.



Supplementary Fig. 5

Expression pattern of the chromosome 14 genes flanking the t(14;21) translocation breakpoint (FBXO33 and LRFN5), and selected genes from the 2q31 microdeletion (SLC25A12, DLX1 and RAPGEF4). Expression data was taken from the publicly available GNF dataset (<http://biogps.gnf.org>).