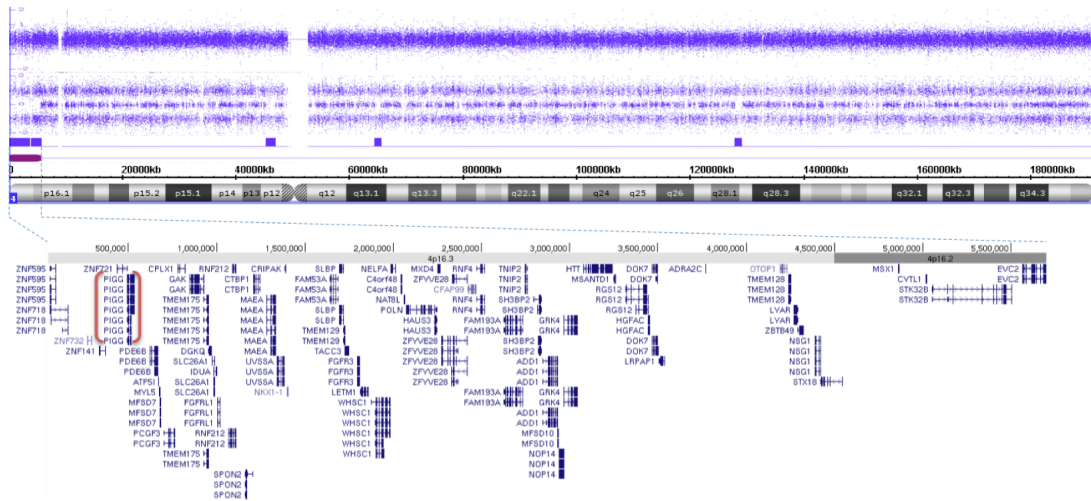


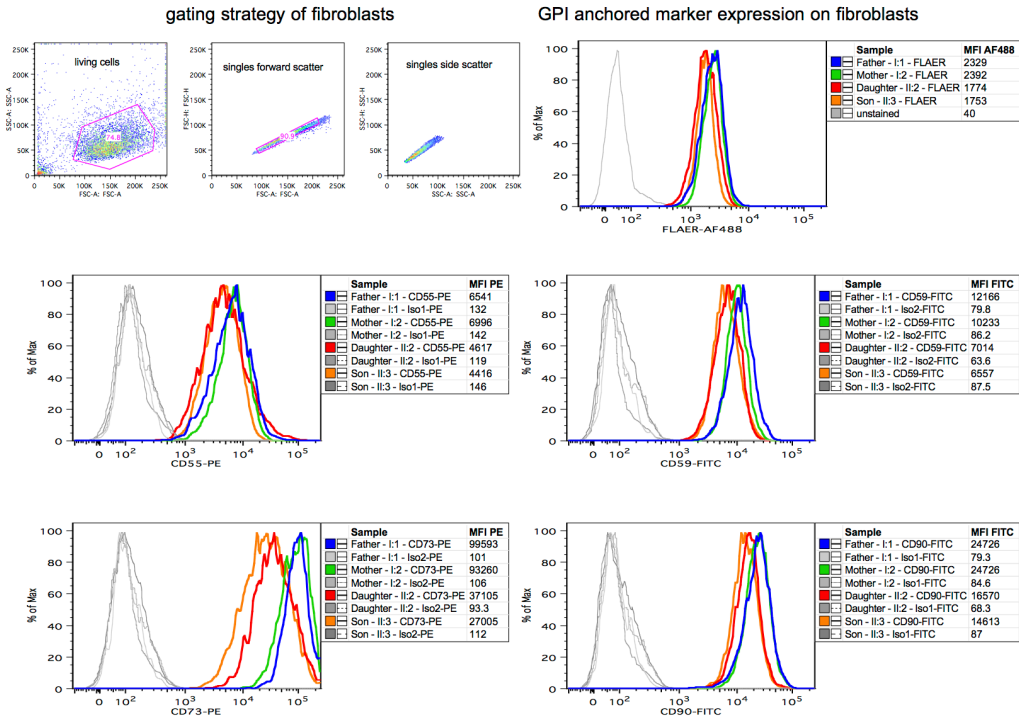
Supplementary Figure S1

MRI image of (A, B) II:2 at age 5 years and 9 months and (C, D) II:3 at age 2 years and 9 months showing cerebellar atrophy with prominent sulci, atrophic cortex and white matter loss.



Supplementary Figure S2

Chromosomal microarray results from patient II:2 of the detected homozygous region at 4p16.3p16.2, with a focused view of the genes encompassed by the homozygous stretch (adapted from UCSC genome browser, assembly GRCh37/hg19).

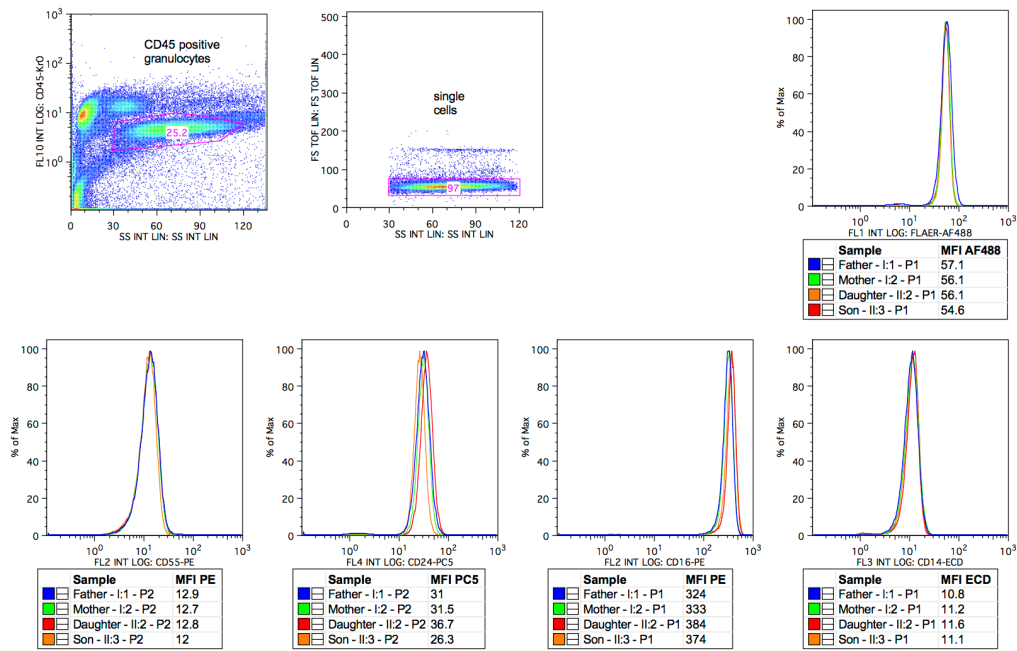


Supplementary Figure S3A

Flow cytometry gating strategy for single fibroblasts and histograms representing median fluorescence intensity (MFI) of GPI anchored marker expression. Patient fibroblasts and control fibroblasts were stained with anti-CD55, anti-CD59, anti-CD73, anti-CD90, and fluorescence-labeled aerolysin (FLAER).

gating strategy for granulocytes

GPI anchored marker expression on granulocytes



Supplementary Figure S3B

Flow cytometry gating strategy for granulocytes from blood samples and histograms representing median fluorescence intensity (MFI) of GPI anchored marker expression