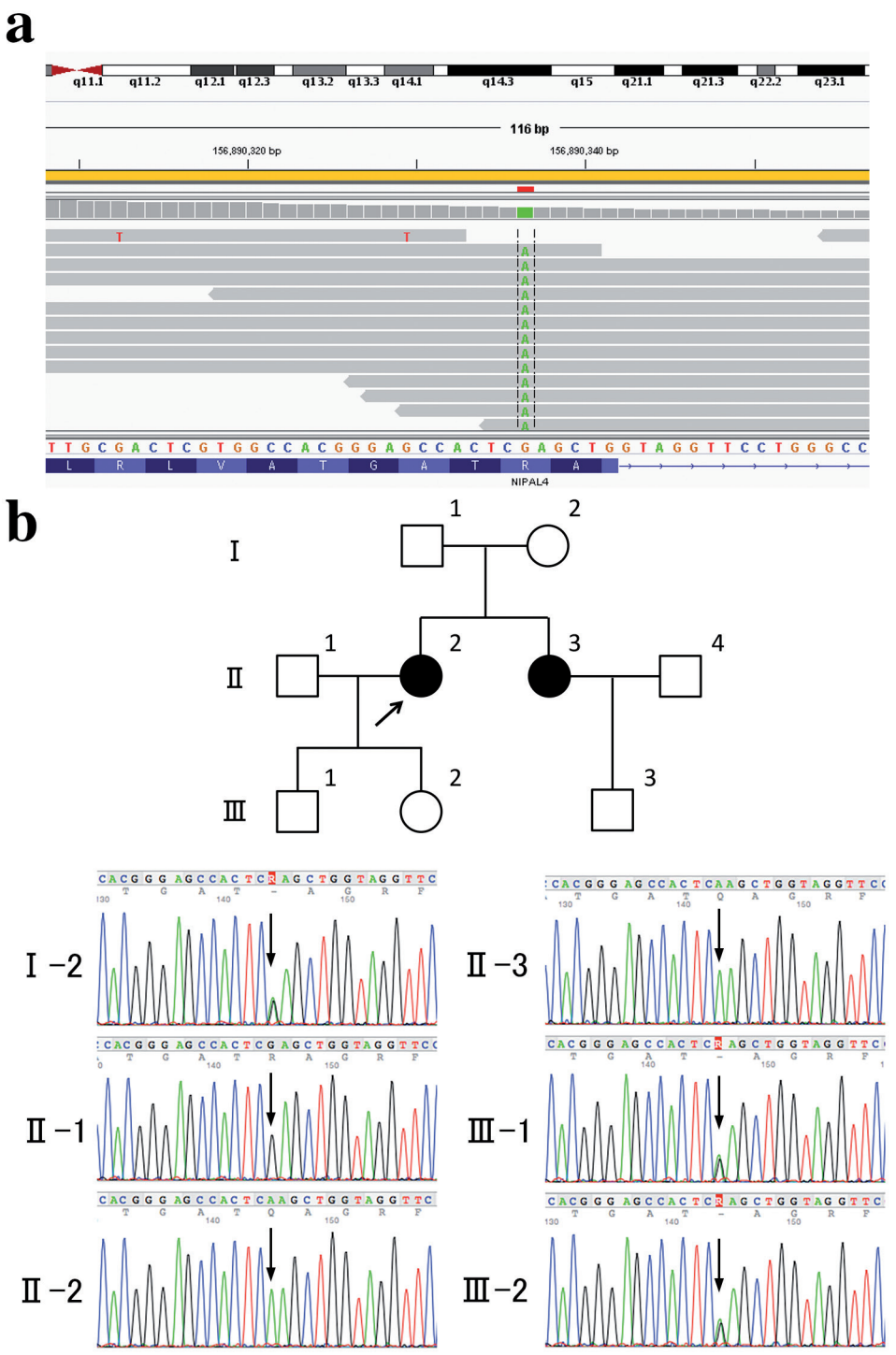

Fig. S2. Next-generation sequencing (NGS) and direct sequencing of the mutation *NIPAL4* c.458G>A in the family. (a) Genome browser view of a homozygous sequence variation chr5:g.[156890336 G>A];[156890336 G>A] in *NIPAL4* identified by NGS. SureSelectXT Human All Exon Kit V6 (Agilent Technologies, Santa Clara, CA, USA) and HiSeq (Illumina, Inc., San Diego, CA, USA) were used for whole-exome sequencing of the patient's genomic DNA. Sequencing data were analysed using Reseq (Amelieff, Tokyo, Japan), StrandNGS (Strand Life Sciences, Pvt. Ltd, Bengaluru, India) and SureCall v.2 (Agilent Technologies). The mean coverage of NGS was 53.43 in QC analysis using StrandNGS. The sequence variation corresponds to a novel homozygous c.458G>A (NM_001099287) in the *NIPAL4* gene. (b) Direct sequencing of the mutation c.458G>A in the family. The arrow indicates the mutated base(s). I-2: c.[458G>A]; [458G=]; II-1: c. [458G=]; [458G=]; II-2: c.[458G>A]; [458G>A]; II-3: c.[458G>A]; [458G>A]; III-1: c.[458G>A]; [458G=]; III-2: c.[458G>A]; [458G=]. R= A and G. The segment of *NIPAL4* genomic DNA was amplified by polymerase chain reaction (PCR) using primers F2976 GTAGCGGAAGCACAGGGTTT and R3648 CGTTGGTGGGAAGTAAGACCA, and was sequenced using the primer S3135 CTCAGCCCTGAGGTGCCC.