SUPPLEMENTARY MATERIAL

Supplementary Figure 1. Genetic findings from the index patient.

A. Electropherograms showing the novel c.767G>A mutation in CCBE1 for the index patient, the patient's parents and siblings. **B.** Family tree showing the distribution of the mutations. The index patient is subject IV.3. Squares represent male family members, and circles female family members. Bi-allelic CCBE1 mutations are reported with black solid symbols, and monoallelic CCBE1 mutations are reported as half-black symbols. Black oblique line represents patients deceased. **C.** Protein domain structure of CCBE1 showing the position of the mutation of the index patient (G256D) (SP: signal peptide; Ca-EGF: Calcium-binding Endothelial Growth Factor). **D.** Ortholog protein sequences alignment: the G in position 256 is highly conserved among species.