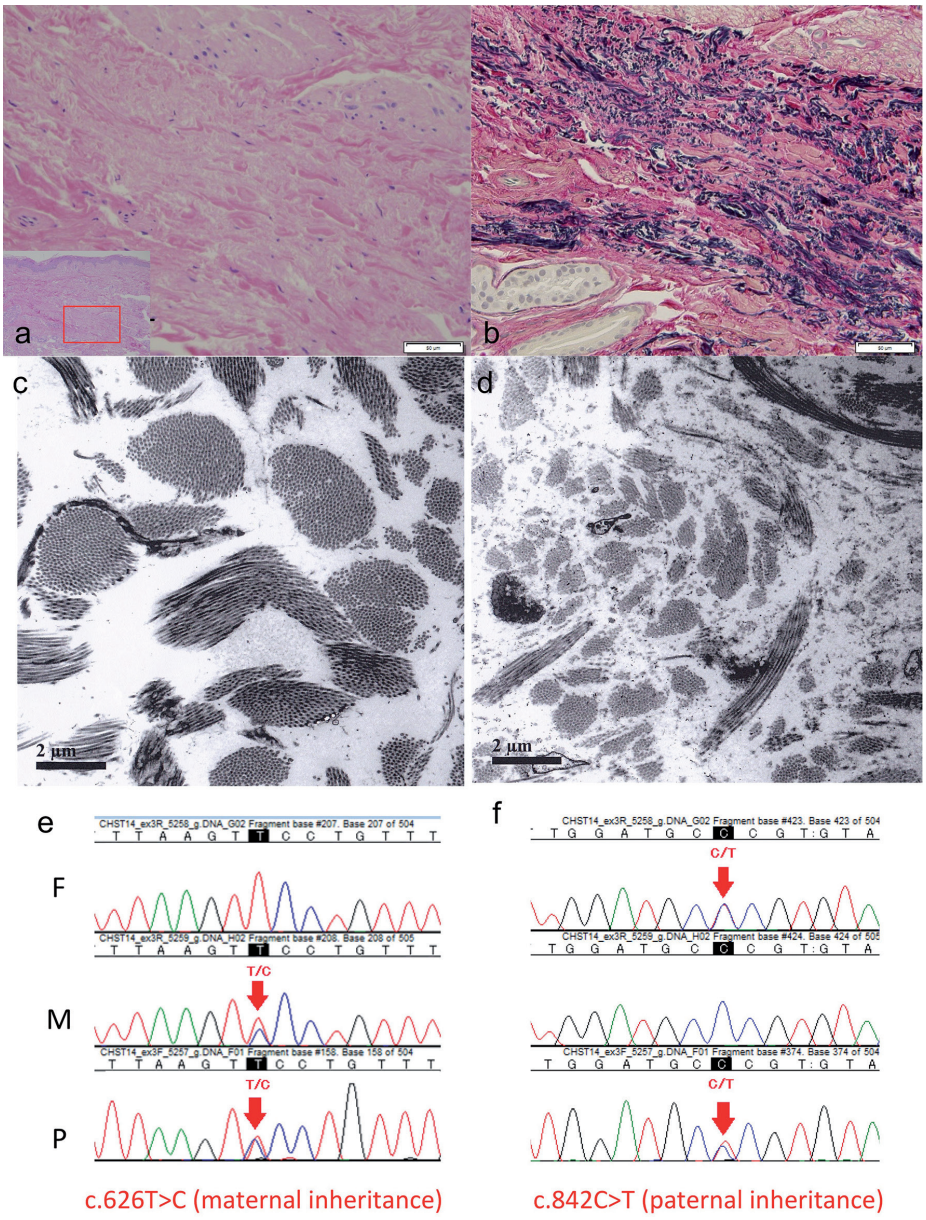

Fig. S2. Pathological examination and molecular investigation. (a, b) Light microscopic findings. A haematoxylin and eosin-stained skin specimen from the patient shows fine collagen fibres to be predominant from the reticular to the papillary dermis, and normally thick collagen bundles in the reticular dermis are markedly reduced (×400) (a). An *elastica* van Gieson-stained skin specimen shows increased elastic fibres in the dermis (×400) (b). (c, d) Electron microscopic findings. Compared with regularly and tightly assembled collagen fibrils in a control specimen (×10,000) (c), the assembly of collagen fibrils is observed to be insufficient in the patient (×10,000) (d). Direct sequencing of *CHST14* on genomic DNA from the patient reveals compound heterozygous mutations (P): c.626T>C (p.Phe209Ser) from her mother (M) and c.842C>T (p.Pro281Leu) from her father (F).