Familial cases of vascular-type Ehlers-Danlos syndrome (vEDS)

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A 38-year-old female was suspected of having vascular-type Ehlers-Danlos syndrome (vEDS) and referred to our department in February 2012. Her skin had bled easily since early childhood, and she had experienced repeated episodes of pneumothorax since 14 years of age. During pregnancy with her first child (at 30 years of age), she developed hydronephrosis on the left side as a result of compression by the uterus. In February 2012, at week 36 of her second pregnancy, cesarean section was performed because of an uterine rupture. Friability of the uterine tissue was noted at the time, and hysterectomy was performed. The patient developed paralytic ileus secondary to a postoperative intra-abdominal hematoma, and because her intestinal tissue was friable, intestinal resection was performed. Cutaneous thinning, easy bleeding, and characteristic facies were noted. We performed an analysis of collagen production by cultured fibroblasts by the method described by Hata et al. and used fibroblasts obtained from healthy skin as a control. The type III/I collagen production ratio was low, only 10.5% of the ratio of production by the control fibroblasts. A genetic analysis of cultured-fibroblast-derived cDNA and blood-derived genomic DNA revealed a missense mutation in exon23 of the COL3A1 gene (c.1538 G>A, p.G→D). At the family’s request we conducted a gene analysis of the COL3A1 gene of the elder son (7 years old) and the younger son (0 years old) and detected the same mutation in the elder son. The elder son has not developed any of the manifestations of vEDS yet, but because of the risk that they will develop as he grows, prevention and very careful follow-up will be necessary.