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# A case of Fabry's disease in a patient with no α-galactosidase A activity caused by a single amino acid substitution of Pro-40 by Ser

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## Abstract

We analyzed a male patient with Fabry's disease who had no activity of the lysosomal hydrolase α-galactosidase A (α-GalA) and female members of his family. We cloned a cDNA that encoded the mutant α-GalA, determined its nucleotide sequence, and found two nucleotide differences between the mutant and the wild-type cDNAs. Although one difference was silent, the other difference, a C-to-T transition at nucleotide number 118, resulted in an amino acid substitution of Pro-40 by Ser. A transient expression assay demonstrated that this missense mutation was the cause of the deficiency of α-GalA activity in the patient. In vitro mutagenesis experiments demonstrated that Pro-40 is critical for the appearance ofa-GalA activity.