Clinical and genetic aspects of Bernard-Soulier syndrome: searching for genotype/phenotype correlations

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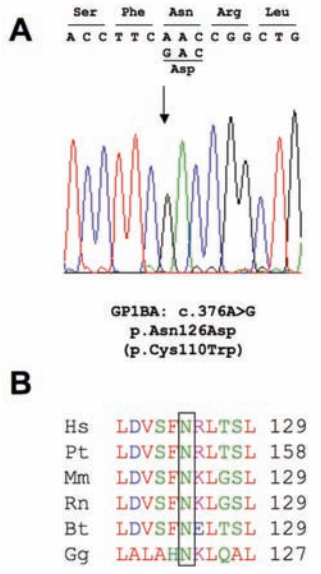

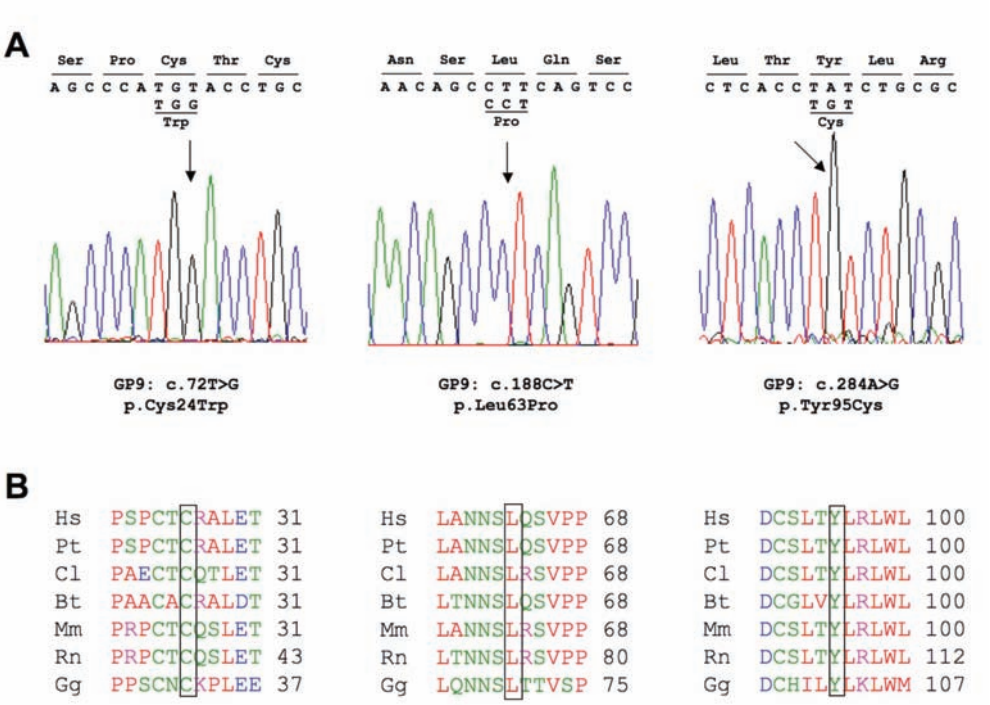

Online Supplementary Figure S1. Characterization of the p.Asn126Asp mutation in *GP1BA*. (A) Electropherogram showing the homozygous p.Asn126Asp mutation identified in family F1. (B) Alignment of GPIbα orthologs from *Homo sapiens* (Hs, NP_000164.4), *Pan troglodytes* (Pt, XP_523557.2), *Mus musculus* (Mm, NP_034456.1), *Rattus norvegicus* (Rn, XP_001080712.1), *Bos Taurus* (Bt, XP_581184.3), and *Gallus gallus* (Gg, XP_418180.2) at *http://www.ncbi.nlm.nih.gov/sites/homologene/143*. The mutated residue is boxed.

Online Supplementary Figure S2. Identification of three novel missense mutations in *GP9*. (A) Direct sequencing analysis of polymerase chain reaction product of *GP9*, showing the homozygous p.Cys24Trp, p.Leu63Pro, and Tyr95Cys mutations identified in families F6, F7 and F9, respectively. (B) Alignment of GPIX orthologs from *Homo sapiens* (Hs, NP_000165.1), *Pan troglodytes* (Pt, XP_001140600.1), *Canis lupus familiaris* (Cl, XP_852017.1), *Bos Taurus* (Bt, XP_875064.2), *Mus musculus* (Mm, NP_061232.1), *Rattus norvegicus* (Rn, NP_001026995.1), and *Gallus gallus* (Gg, XP_414314.1) at *http://www.ncbi.nlm.nih.gov/sites/homologene/144*. The mutated residues are boxed.