A large Swiss family with Bernard-Soulier syndrome - Correlation phenotype and genotype

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Bernard-Soulier syndrome (BSS) is a rare, autosomal recessive inherited bleeding disorder associated with thrombocytopenia, thrombocytopathy and giant platelets. BSS is caused by genetic alterations of the glycoprotein (GP) Ib/V/IX complex. We report on a large Swiss family of whom four family members suffer from BSS. Here, a homozygous missense mutation in position 1829 (A(R)G) of the GPIX gene constituting a N45S substitution is the cause for the bleeding symptoms. A total of 38 family members within two generations were analyzed regarding the N45S mutation by DNA sequencing and restriction fragment length polymorphism. The laboratory parameters which are characteristically for BSS such as platelet count, platelet volume and the expression of CD42a (GPIX), CD42b (GPIbalpha) and CD41 (GPIIb) were measured for all 38 individuals. The four homozygous patients showed bleeding symptoms, thrombocytopenia and giant platelets. In these patients, the expression of CD42a (GPIX), CD42b (GPIbalpha) was diminished. Interestingly, the intensity of the bleeding symptoms of the 4 homozygous family members seemed to vary although they carry the same mutation. The 24 heterozygous carriers did not differ significantly from their 10 wildtype family members regarding bleeding symptoms and laboratory analysis.
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