

Platelet functional abnormalities and clinical presentation in pediatric patients with germline *RUNX1*, *ANKRD26*, and *ETV6* mutations

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SUPPLEMENTARY MATERIALS

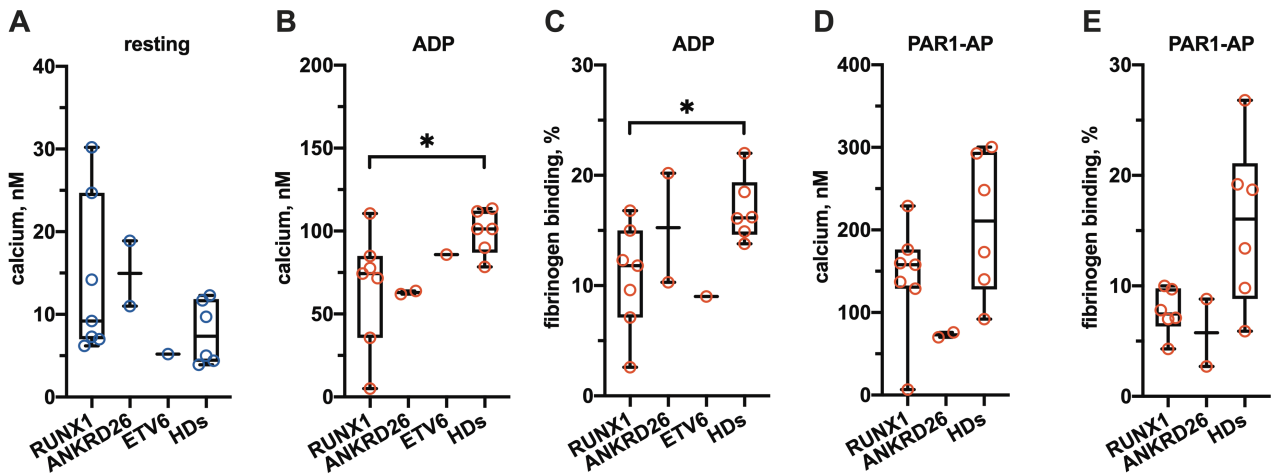


Figure S1. Platelet flow cytometry (Platelet signaling study) in patients with *RUNX1*, *ANKRD26*, *ETV6* mutations, and healthy children. The data points are the circle symbols, horizontal lines are medians, boxes show 25th–75th percentiles, error bars show 5–95% intervals. Blue color refers to resting platelets, red color refers to stimulated platelets. Statistical significance was calculated using Mann-Whitney criteria, * corresponds to $p < 0.05$; no marking corresponds to non-significant differences. A – Cytosolic calcium concentration in resting platelets is increased in patients with *RUNX1* and *ANKRD26* mutations, while it is normal in the patient with *ETV6* mutation. B, C – Platelet calcium responses (B) and fibrinogen binding (C) upon stimulation with 2 μM ADP are predominantly diminished in patients with inherited thrombocytopenias. D, E – Cytosolic calcium mobilization (D) and fibrinogen binding (E) in the response to 5 μM PAR1-AP are also decreased in most of the patients.

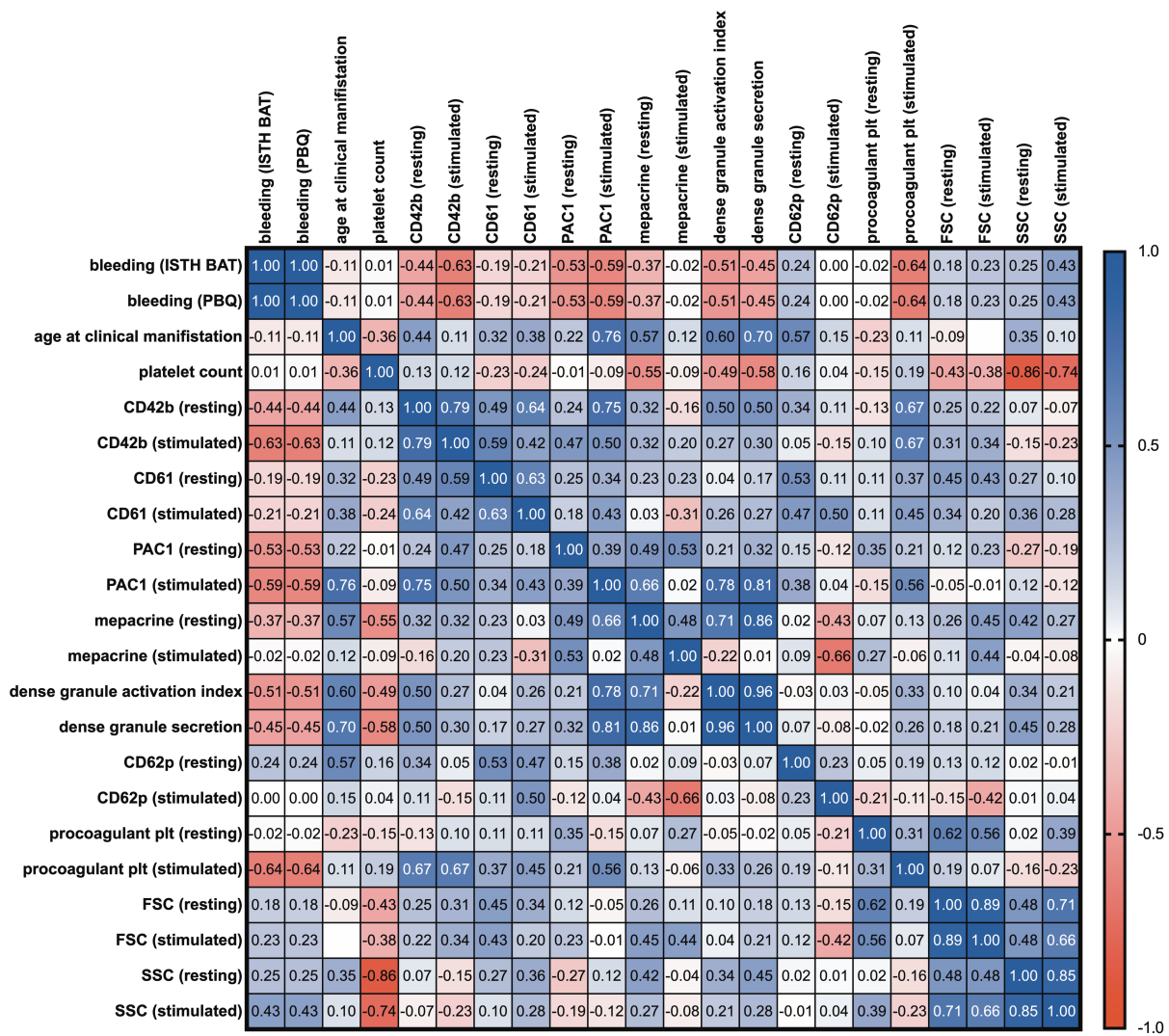


Figure S2. Correlations between clinical and laboratory data in patients with germline *RUNXI* mutations (Spearman r)

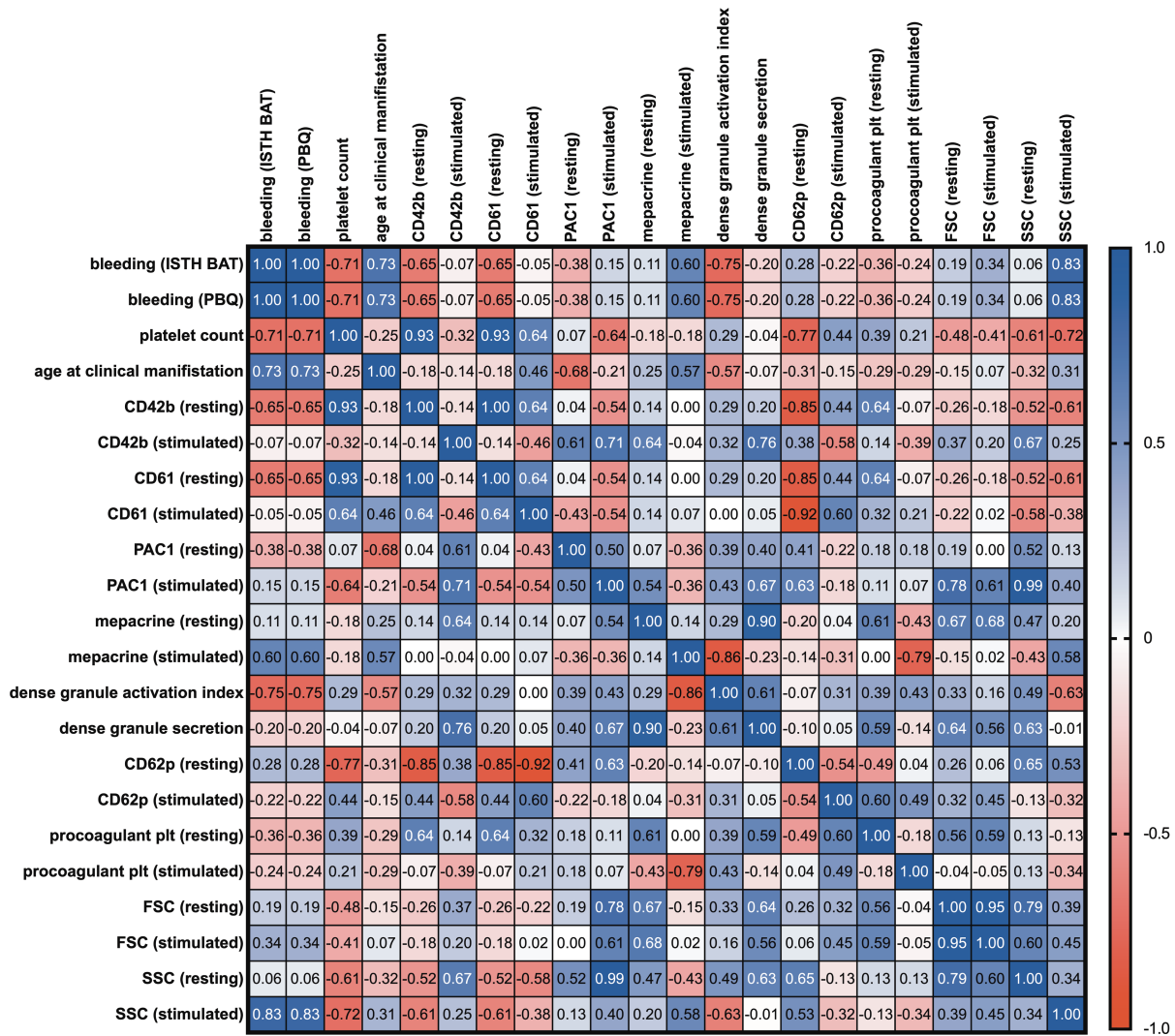


Figure S3. Correlations between clinical and laboratory data in patients with germline ANKRD26 mutations (Spearman r)