

Supplementary material

Table S1. Genetics and clinical features of SDS patients enrolled in this study.

UPN	Gender	Age	Genotype	PMN (Cell/mm ³)	Phenotype	Cytogenetics
20	M	46	258+2T>C/183-184TA>CT	500	PI, FTT, bone malformation, thrombocytopenia	46,XY, del(20)q
26	M	16	258+2T>C/183-184TA>CT	58	PI, FTT, bone malformation, thrombocytopenia	46,XY
37	F	10	258+2T>C/183-184TA>CT	1280	PI, FTT, recurrent infections	46,XX i(7)(q10)
39	M	41	258+2T>C/183-184TA>CT	1300	PI, FTT, bone malformation, thrombocytopenia, cognitive impairment	46,XY
47	M	12	258+2T>C/183-184TA>CT	550	PI, FTT, recurrent infections, bone malformation, thrombocytopenia	46,XY
58	M	12	258+2T>C/183-184TA>CT	390	PI, FTT, HbF>2%, bone malformation, thrombocytopenia, anemia	46,XY
69	F	8	258+2T>C/183-184TA>CT	770	PI, FTT, HbF>2%, anemia, cognitive impairment	46,XX
72	M	28	258+2T>C/183-184TA>CT	380	PI, FTT, recurrent infections, HbF>2%, bone malformation, thrombocytopenia, anemia, cognitive impairment	46,XY
73	F	7	258+2T>C/183-184TA>CT	520	PI, FTT, HbF>2%, thrombocytopenia	46,XX
74	M	9	258+2T>C/183-184TA>CT	1430	PI, FTT, HbF>2%, cognitive impairment	46,XY
75	F	7	258+2T>C/183-184TA>CT	1000	PI, FTT, HbF>2%, bone malformation, thrombocytopenia, cognitive impairment	46,XX
82	M	8	258+2T>C/183-184TA>CT	310	PI, FTT, recurrent infections, bone malformation, thrombocytopenia, anemia, cognitive impairment	46,XY
80	M	7	258+2T>C/183-184TA>CT	680	PI, FTT, recurrent infections, bone malformation, anemia, cognitive impairment	46,XY
87	M	18	258+2T>C/183-184TA>CT	880	PI, FTT, recurrent infections, bone malformation, cognitive impairment	46,XY
106	M	36	258+2T>C/183-184TA>CT	1210	PI, FTT, bone malformation, anemia	46,XY
108	M	17	258+2T>C/183-184TA>CT	970	PS, FTT, bone malformation, anemia	46,XY

UPN, unique patient number; PI, pancreas insufficiency; PS, pancreas sufficiency; FTT, failure to thrive.