

Supplementary Tables

Chrom	Position	Gene	Refseq	Protein variant	Type of variant	Genotype
chr1	6195297	CHD5	NM_015557	M955V	missense	C/T
chr1	145560247	ANKRD35	NM_144698	R245W	missense	C/T
chr1	222717489	HHIPL2	NM_024746	T122A	missense	C/T
chr5	137589524	GFRA3	NM_001496	R319*	nonsense	A/G
chr8	48508474	SPIDR	NM_001080394	V400A	missense	C/T
chr8	90983508	NBN	NM_002485	P199S	missense	A/G
chr9	114338650	ZNF483	NM_001007169	H243Y	missense	C/T
chr11	85445332	SYTL2	NM_032943	G346E	missense	C/T
chr11	125851006	CDON	NM_001243597	G1072R	missense	C/T
chr12	122406044	WDR66	NM_144668	R914C	missense	C/T
chr16	19873179	GPRC5B	NM_016235	V383I	missense	C/T
chr16	48121966	ABCC12	NM_033226	R1169H	missense	C/T
chr16	83781822	CDH13	NM_001220488	V597M	missense	A/G
chr17	7591983	WRAP53	NM_018081		exon	CT/-
chr17	40269120	KAT2A	NM_021078	R566H	missense	C/T
chr19	48385433	SULT2A1	NM_003167	L127S	missense	A/G
chr22	37603284	SSTR3	NM_001278687	G187R	missense	C/G
chrX	114425490	RBMXL3	NM_001145346	D496N	missense	A/G

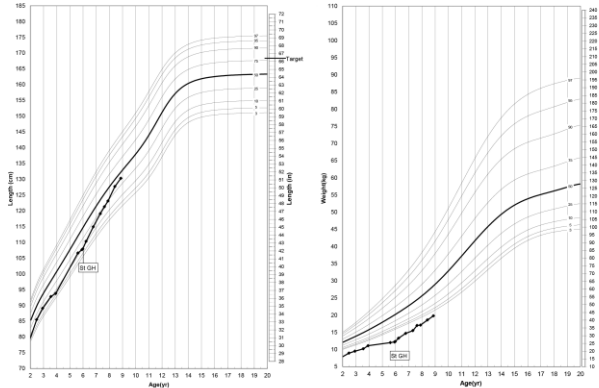
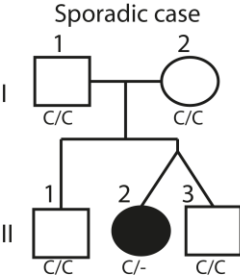
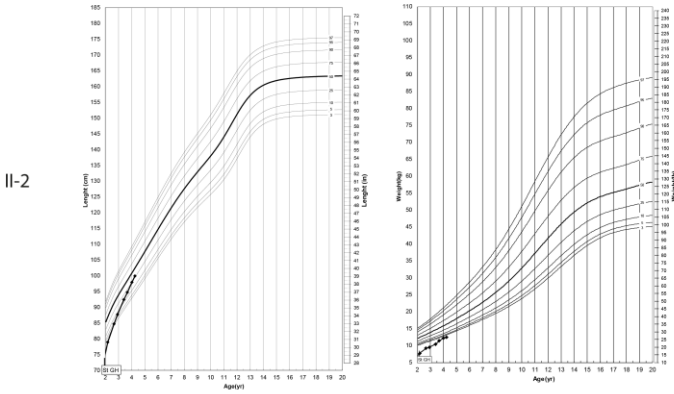
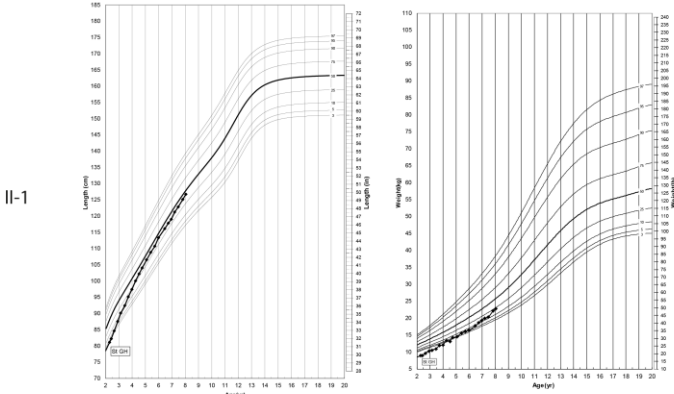
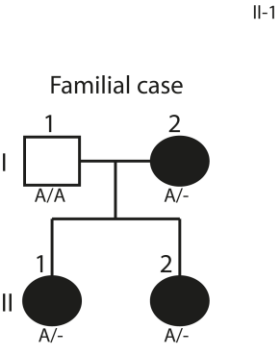
Supplementary Table 1: These 18 other variants found in the affected members of the family have no known effect on growth. These variants were not validated by Sanger sequencing.

GAPDH-F	GAAGGTGAAGGTCGGAGTCAAC
GAPDH-R	CAGAGTTAAAAGCAGCCCTGGT
total-IGF2-F	TGGACACCCTCCAGTTCGTC
total-IGF2-R	GCGGAAACAGCACTCCTCAA
IGF2-P3-F	GTCCCCACAGACGAACTGGA
IGF2-P3-R	TTCGGTTTGCGACACGCAG
IGF2-P1-F	CTGCTGGAGCTGAGCCTCAG
IGF2-P1-R	TGAGCTGGCAGCGATTCAG
H19-F	GGTGGCCATGAAGATGGAGT
H19-R	GTGTGACGGCGAGGACAG

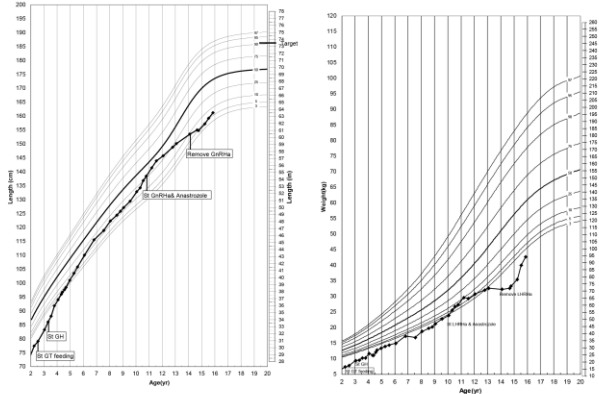
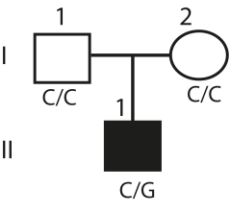
Supplementary Table 2: Sequence of the RT-qPCR primers used in this study.

Supplementary Figure

PLAG1 mutations



IGF2 mutation NM_000612.5:c.78C>G



Pedigrees, growth charts (2 to 20 years of age) of patients with SRS carrying mutations of the *PLAG1* and *IGF2* genes.