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Supplemental Data

Exome Sequencing Identifies Autosomal-Dominant *SRP72* Mutations Associated with Familial Aplasia and Myelodysplasia

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Table S1. The 107 Exome Variations Shared by All Affected Members of Family 1

Chr	Nucleotide Position	Reference Base	Sample Base	Variation Effect	AA WT	AA Var	Protein Position	Gene	MIM	Follow-Up
1	12776677	T	A	nonsense	LEU	stop	105/475	*PRAMEF1*	na	NL
1	12923773	C	T	missense	CYS	TYR	166/477	*PRAMEF6*	na	NL
1	12959174	C	T	missense	PRO	LEU	220/482	*PRAMEF22*	na	NL
1	52931112	A	C	missense	VAL	GLY	41/232	*C1orf163*	na	NL
1	108095298	C	T	missense	ARG	GLN	408/848	*VAV3*	605541	EVS
1	143663577	C	T	missense	ALA	THR	167/174	*PDE4DIP*	608117	NL
1	150543001	C	G	missense	SER	THR	3662/4062	*FLG*	135940	NL
1	150543083	C	T	missense	ASP	ASN	3635/4062	*FLG*	135940	NL
1	150544178	G	A	missense	ARG	CYS	3270/4062	*FLG*	135940	NL
1	150545179	T	C	missense	ASP	GLY	2936/4062	*FLG*	135940	NL
1	150546353	C	T	missense	GLY	ARG	2545/4062	*FLG*	135940	NL
1	150546656	T	C	missense	LYS	GLU	2444/4062	*FLG*	135940	NL
1	150546794	C	G	missense	GLU	GLN	2398/4062	*FLG*	135940	NL
1	150547095	C	G	missense	GLU	ASP	2297/4062	*FLG*	135940	NL
1	150547941	G	T	missense	ASP	GLU	2015/4062	*FLG*	135940	NL
1	150548369	G	T	missense	GLN	LYS	1873/4062	*FLG*	135940	NL
1	150549534	G	C	missense	ASP	GLU	1484/4062	*FLG*	135940	NL
1	150551723	C	T	missense	GLU	LYS	755/4062	*FLG*	135940	NL
1	150551805	G	T	missense	HIS	GLN	727/4062	*FLG*	135940	NL
1	151066748	G	T	missense	GLY	VAL	59/111	*LCE1A*	612603	NL
1	156070884	G	A	missense	PRO	SER	219/348	*CD5L*	602592	NL
1	156072297	C	A	nonsense	GLU	stop	110/348	*CD5L*	602592	NL
1	184543711	G	C	missense	CYS	SER	746/1405	*PRG4*	604283	NL
1	246291192	G	T	missense	GLY	CYS	196/313	*OR2L13*	na	NL
1	246703738	T	C	missense	VAL	ALA	155/319	*OR2T3*	na	NL
1	246704166	C	T	missense	ARG	CYS	298/319	*OR2T3*	na	NL
1	246803882	C	G	missense	ARG	PRO	267/319	*OR2T34*	na	NL
1	246868215	C	T	missense	GLY	ASP	323/324	*OR2T35*	na	NL

Chr	Nucleotide Position	Reference Base	Sample Base	Variation Effect	AA WT	AA Var	Protein Position	Gene	MIM	Follow-Up
2	108642531	G	A	missense	ARG	HIS	12/326	*LIMS1*	602567	NL
2	112330900	G	A	nonsense	GLN	stop	465/1945	*ANAPC1*	608473	NL
2	130616274	T	C	missense	HIS	ARG	149/381	*CCDC74B*	na	NL
2	135606966	C	G	missense	PRO	ARG	423/982	*RAB3GAP1*	602536	REPORT
2	140830464	G	A	missense	ALA	VAL	3816/4600	*LRP1B*	608766	EVS
2	160312787	G	A	missense	SER	ASN	247/705	*March7*	613334	NL
2	160398902	G	A	missense	PRO	LEU	1247/1723	*LY75*	604524	NL
4	56968604	C	G	missense	GLY	ALA	55/518	*PPAT*	172450	EVS
4	57,045,760	CA	del	indel	THR	LYSfs*19	355/671	*SRP72*	602122	REPORT
5	118530264	G	T	missense	GLU	ASP	1675/3028	*DMXL1*	605671	EVS
5	140210717	G	A	missense	ARG	GLN	818/843	*PCDHA9*	606315	EVS
5	177352572	G	A	missense	ALA	VAL	142/227	*PROP1*	601538	EVS
6	150384904	A	G	missense	MET	THR	85/247	*RAET1L*	611047	EVS
7	5950079	A	C	missense	PHE	CYS	533/871	*RSPH10B*	na	NL
7	73850247	G	T	missense	HIS	ASN	514/950	*GTF2IRD2*	608899	EVS
7	74983431	G	A	missense	ARG	TRP	23/110	*PMS2L3*	na	NL
7	75964673	C	T	missense	ARG	CYS	365/623	*DTX2*	613141	NL
7	143047878	C	T	missense	ARG	TRP	265/846	*FAM115C*	na	NL
7	143048016	T	C	missense	CYS	ARG	311/846	*FAM115C*	na	NL
7	151557958	A	G	missense	TYR	HIS	987/4912	*MLL3*	606833	NL
7	151575940	C	T	missense	GLY	SER	838/4912	*MLL3*	606833	NL
8	11452901	A	C	missense	LYS	THR	325/506	*BLK*	191305	EVS
8	17146938	C	T	missense	ARG	GLN	32/286	*CNOT7*	604913	EVS
9	20771801	A	G	missense	GLN	ARG	357/1802	*KIAA1797*	na	NL
9	33375852	C	T	missense	GLY	ARG	180/343	*AQP7*	602974	EVS
9	39167397	G	A	missense	THR	MET	282/1289	*CNTNAP3*	610517	EVS
9	67558278	G	A	missense	ALA	THR	673/824	*ANKRD20A1*	na	EVS
9	67558296	C	T	missense	ARG	CYS	679/824	*ANKRD20A1*	na	DNS
9	68496744	T	A	missense	GLU	ASP	288/396	*CBWD6*	na	EVS
9	68713575	G	C	missense	SER	THR	684/824	*ANKRD20A4*	na	NL
9	71044679	C	T	missense	HIS	TYR	788/1191	*TJP2*	607709	DNS
9	95137568	C	T	missense	GLY	SER	92/197	*C9orf129*	na	NL
10	51135558	A	T	missense	TYR	ASN	302/664	*CTGLF4*	na	DNS
10	102047352	A	C	missense	TRP	GLY	245/806	*PKD2L1*	604532	EVS
10	105174891	G	A	missense	GLY	ASP	975/1872	*PDCD11*	612333	NL
11	18224054	G	A	missense	ALA	VAL	70/123	*SAA2*	104751	EVS
11	48303492	G	C	missense	VAL	LEU	142/330	*OR4C3*	na	NL
11	48303508	G	A	missense	ARG	HIS	147/330	*OR4C3*	na	NL
11	48303537	A	T	missense	ASN	TYR	157/330	*OR4C3*	na	NL
11	48303538	A	G	missense	ASN	SER	157/330	*OR4C3*	na	NL
12	11397478	A	C	missense	SER	ALA	276/332	*PRB1*	180989	EVS
12	14834818	G	A	missense	THR	ILE	383/642	*WBP11*	na	NL
12	121411651	C	A	missense	LEU	PHE	271/1428	*CLIP1*	179838	NL
13	23919263	T	C	missense	GLN	ARG	1059/1725	*PARP4*	607519	NL
13	23919323	A	G	missense	ILE	THR	1039/1725	*PARP4*	607519	NL
14	18448030	T	A	missense	LEU	HIS	146/327	*OR11H12*	na	EVS
15	37850100	C	T	missense	ASP	ASN	44/582	*FSIP1*	na	EVS
15	38978455	G	A	missense	ALA	THR	98/974	*VPS18*	608551	EVS
15	40226736	C	T	missense	ALA	THR	433/850	*PLA2G4F*	na	EVS
16	24712455	A	T	missense	GLN	HIS	1112/1963	*TNRC6A*	610739	NL

Chr	Nucleotide Position	Reference Base	Sample Base	Variation Effect	AA WT	AA Var	Protein Position	Gene	MIM	Follow-Up
17	8099890	G	A	missense	ARG	HIS	206/1339	*PFAS*	602133	NL
17	21259375	G	A	missense	ARG	HIS	43/434	*KCNJ12*	602323	NL
17	21259460	G	A	missense	MET	ILE	71/434	*KCNJ12*	602323	NL
17	21259662	G	A	missense	GLU	LYS	139/434	*KCNJ12*	602323	NL
17	21259680	G	A	missense	GLY	SER	145/434	*KCNJ12*	602323	NL
17	21259764	G	A	missense	ASP	ASN	173/434	*KCNJ12*	602323	NL
17	21259801	C	T	missense	ALA	VAL	185/434	*KCNJ12*	602323	NL
17	21259878	C	T	missense	LEU	PHE	211/434	*KCNJ12*	602323	NL
17	21260136	G	A	missense	VAL	ILE	297/434	*KCNJ12*	602323	NL
17	21260153	G	T	missense	MET	ILE	302/434	*KCNJ12*	602323	NL
17	41981976	C	G	missense	ASN	LYS	1385/1701	*LRRC37A2*	na	EVS
17	43175021	A	C	missense	THR	PRO	180/536	*TBX21*	604895	NL
17	60322733	A	T	missense	SER	THR	369/1635	*LRRC37A3*	na	EVS
19	38182406	T	C	missense	GLN	ARG	384/687	*RHPN2*	612591	NL
19	42334762	C	T	missense	VAL	ILE	572/715	*ZNF585A*	na	EVS
19	45068502	G	C	missense	HIS	GLN	3920/5406	*FCGBP*	na	EVS
19	45084425	T	G	missense	GLU	ALA	2640/5406	*FCGBP*	na	EVS
19	54346373	C	G	missense	GLU	GLN	698/700	*HRC*	142705	EVS
19	55985910	C	T	missense	PRO	LEU	95/427	*ACPT*	606362	EVS
19	59563476	G	C	missense	PRO	ARG	127/288	*LAIR1*	602992	EVS
19	61012109	G	A	missense	THR	MET	560/1034	*NLRP11*	609664	NL
19	62338579	C	T	missense	GLY	GLU	313/473	*ZIM3*	na	EVS
19	63062058	T	C	missense	SER	PRO	156/576	*ZNF587*	na	EVS
22	23445729	A	G	missense	TYR	HIS	840/883	*PIWIL3*	610314	REPORT
X	2149200	T	C	missense	HIS	ARG	292/331	*DHRSX*	na	X-chr
X	2171129	C	G	missense	VAL	LEU	247/331	*DHRSX*	na	X-chr
X	7772017	C	T	missense	PRO	LEU	194/207	*VCX*	300533	X-chr
X	140821571	C	T	missense	PRO	SER	239/1143	*MAGEC1*	300223	X-chr
X	153071718	C	T	missense	ALA	VAL	174/365	*OPN1LW*	300822	X-chr

Chr – chromosome; AA WT – wild type amino acid. AA var – variant amino acid; Nucleotide position – base affected relative to hg18 build; na – no entry on OMIM; Follow up - the criteria used to follow or exclude this mutation from further analysis: NL – not linked (mutation lies outside the linkage region); EVS – mutation reported on Exome Variant Server database; DNS – mutation does not segregate with disease; REPORT – detailed in this paper.; X-chr – mutation not followed up as on X chromosome.

Table S2. Primer Sequences Used in this Paper

Gene	Exon/Mutation	Forward Primer	Reverse Primer
SRP72	1	GAT CTC TGA TGC TGC CTT AG	CCG GGC GGA CAA TAG AC
SRP72	2	TAG CTT TCG GAG AGA CAG G	TTC CAG TTC AGA AAC ATT ATT GAC
SRP72	3	GCT AGG GAA GTT GAT ATA TGC	GGA CTG GTT TGG TGG TCA G
SRP72	4+5	GTG GTT CCA AAA AAG GTT TGC	TGT CAC TGC ATC AAT AAC TTC C
SRP72	6 / c.620G>A	AGC AAA GTG CCA AAG AGC TG	GGT GGT TTC TAG TGT GAT TCC
SRP72	7+8	TTG TAT CTC TTT TGA ATG TTT GGG	TGT GAT TTT GCT ACA ACT GGA C
SRP72	9	GCT TAG GCA CCC CAA TCA G	TAC AGG CAT GAG CCA TTG TG
SRP72	10 / c.1064_1065del	TAC CCT AGG CAG TTC TTT GG	GTA CAT TTC TGA TTA GTT TTC CC
SRP72	11	GCA CTT ACT TTT AGA GCA TAC C	TCT AGC TGC ATG TTT GAA ACA G
SRP72	12	GGA AAA GTA GTG GGA GGA AG	GAA ACC AAT ACC AGA GCA GAG
SRP72	13	ATC AGG TAT TAG CGG AGA GC	TGA CGT GTA ATA CTT GTT TGC C

Gene	Exon/Mutation	Forward Primer	Reverse Primer
SRP72	14+15	CAT AGG TGT ATT TCT TAA AGG TC	AAA AGC TCT GGT CAA AAC ACA G
SRP72	16	CAG TGT CAA TAG ATA CAT TGG C	GGT TAA TTT TGG GCT GAG GG
SRP72	17	CCA TGT TTA TGA TAA GTG CCC	AAA GAT ACA AGG GAG GGC AG
SRP72	18	TCC CAG GCT TAG AGA CAT C	CAA AGA TAC ATC ACA TTT CTC ATA C
SRP72	19	TGT GGG AGA TTG TTA AGT GTG	GTT CTG TGC TTC TTG CTG TG
PIWIL3	c.2518T>C	CAC GAG AGT TAC TGT GTC AG	TAG AAG CCA GTT CAG AGG TC
RAB3GAP1	c.1268C>G	TGT AGA ACA AAC TGG CCA ATA C	GAT CTG CCA TCC AAA ATT CCC