

Additional File 2. SNPs identified through exome sequencing

	ND02537 (proband)		ND02538 (mother)		ND02539 (Father)		ND02540 (sister)	
DNA source	PBMC	LCL	PBMC	LCL	PBMC	LCL	PBMC	LCL
Total SNPs (novel)	18660 (485)	18794 (491)	16184 (388)	16316 (401)	17531 (467)	17597 (471)	18079 (617)	18216 (574)
Shared SNPs	18433		15963		17214		17806	
Variants unique to sample	227	361	221	353	317	383	273	410
SNP concordance rate between DNA sources	96.81%		96.40%		95.93%		96.16%	
Total non-coding SNPs (novel)	8599 (145)	8725 (150)	7486 (175)	7558 (190)	7710 (201)	7746 (170)	7489 (293)	7694 (267)
% of SNPs in non-coding regions	46.08%	46.42%	46.26%	46.51%	43.98%	44.02%	41.42%	42.24%
Shared SNPs	7486		6392		7508		7439	
SNPs unique to sample	113	239	94	166	202	238	50	255
Total cSNPs (novel)	10061 (340)	10069 (341)	8698 (213)	8758 (211)	9821 (266)	9851 (301)	10590 (324)	10522 (307)
% of SNPs in coding regions	53.92%	53.58%	53.69%	53.68%	56.02%	55.98%	58.58%	57.76%
Shared SNPs	9947		8571		9706		10367	
Discordant SNPs (novel)	114 (8)	122 (9)	127 (11)	187 (9)	115 (2)	145 (37)	223 (28)	155 (11)
Concordant SNPs								
Synonymous (novel)	5036 (119)		4184 (64)		4955 (97)		5786 (114)	
Non-synonymous (novel)	4220 (193)		3772 (115)		4076 (150)		4879 (165)	
MicroRNA (novel)	5		4		5 (1)		2	
Near Gene (novel)	143 (5)		154 (5)		168 (5)		168 (2)	
Nonsense (novel)	40 (6)		30 (5)		30 (4)		45 (3)	
Splice (novel)	7		3		7		8 (1)	
UTR (novel)	488 (9)		424 (13)		46 (7)		479 (11)	
Discordant SNPs								
Synonymous (novel)	61 (3)	60 (5)	69 (5)	135 (4)	35 (2)	76 (13)	87	42 (2)
Non-synonymous (novel)	57 (5)	54 (4)	36 (5)	50 (5)	78	64 (22)	135 (28)	111 (9)
MicroRNA					1	1	1	2
Near Gene	2		8					
Nonsense (novel)			(1)					
Splice								
UTR	2						4 (2)	

Novel SNP: not represented in either dbSNP or 1000 Genomes Project Data

Near Gene: SNP located within 50 bp 3'- or 5'- of a gene