SUPPLEMENTAL MATERIAL

Byun et al., http://www.jem.org/cgi/content/full/jem.20130592/DC1

Table S1. Genomic regions with positive LOD scores identified by homozygosity mapping

Chromosome	Size	Number of SNPs	Max. LOD score	5′ SNP	5′ SNP position (GRCh37)	3′ SNP	3′ SNP position (GRCh37)
	Mb						
1	1.36819	10	1.3132	rs11240777	798959	rs262680	2167149
1	1.938975	93	1.3285	rs2480054	14792535	rs525409	16731510
1	5.639601	334	1.3285	rs2143808	17282425	rs209693	22922026
1	0.348616	4	0.5198	rs10902682	28274984	rs6669686	28623600
1	17.196349	345	1.3285	rs12118994	65029454	rs9324187	82225803
1	2.174524	50	1.3285	rs10493711	82689732	rs3738573	84864256
1	2.675807	57	1.3285	rs7514763	84930166	rs10873820	87605973
1	0.83033	5	0.7945	rs4601565	192621857	rs1410511	193452187
2	0.615297	16	1.2718	rs4240225	19013114	rs13012083	19628411
2	0.360373	12	1.2828	rs4422109	142165632	rs10170191	142526005
2	0.525667	14	1.2859	rs887701	191562927	rs1551439	192088594
2	1.155207	11	0.5656	rs6727480	198143871	rs11688831	199299078
3	9.80742	309	1.3285	rs2254295	8802292	rs17044638	18609712
3	1.849264	71	1.3285	rs10936674	170931099	rs9833380	172780363
4	0.495594	13	1.0414	rs4699738	100175530	rs1426730	100671124
5	1.748478	16	1.3166	rs17160705	100202927	rs1505420	101951405
6	1.565978	53	1.3285	rs3019442	162170524	rs6903758	163736502
7	1.933765	60	1.3285	rs992586	14210392	rs10950603	16144157
7	0.197451	6	0.6458	rs1362921	33914209	rs961652	34111660
8	0.23235	10	0.6397	rs4354280	98022432	rs6982771	98254782
9	2.176653	72	1.3285	rs10816043	9064648	rs901992	11241301
9	0.112562	5	0.9729	rs2102121	107815682	rs7024094	107928244
10	2.85421	136	1.3285	rs7079384	2692656	rs12258676	5546866
10	0.113017	6	0.0244	rs2243668	33563893	rs2776938	33676910
11	3.469697	50	1.3174	rs4755435	36136580	rs4315042	39606277
11	7.09564	128	1.3285	rs11035461	39799738	rs11039014	46895378
12	5.046596	216	1.3285	rs11063263	191619	rs501004	5238215
13	2.299283	51	1.3285	rs1216860	47232268	rs7338291	49531551
13	9.038625	239	1.3285	rs9302065	95971016	rs9519304	105009641
14	0.755496	27	1.3276	rs3811209	22969651	rs977870	23725147
14	0.155317	5	0.7288	rs17105565	37335558	rs35576618	37490875
14	0.22659	4	1.2251	rs1016684	58206045	rs17094025	58432635
15	6.65889	317	1.3285	rs8043364	92013902	rs12441557	98672792
17	0.374185	5	0.6798	rs9748016	410451	rs9892880	784636
17	1.484395	24	1.2836	rs886076	49839893	rs1607977	51324288
17	0.660638	22	1.3285	rs12949090	78217097	rs868432	78877735
18	0.789182	16	1.3228	rs11662562	32232892	rs355317	33022074

Table S2. List of variations unique to the patient that are homozygous and within the intervals identified by genome-wide linkage

Gene	Chromosome	Position (GRCh37)	Reference base	Sample base	Consequence	PolyPhen-2	SIFT
TNFRSF4	1	1149118	G	A	p.R65C	Probably damaging	Damaging
CLCNKA	1	16360484	C	G	Utr	NA	NA
IQSEC1	3	12940954	G	A	Utr	NA	NA
NR2C2	3	15093941	G	A	Utr	NA	NA
OXA1L	14	23240510	G	A	p.R363Q	Benign	Tolerated

NA, not applicable.