Supplementary Data

Homozygosity Mapping and Whole Exome Sequencing to Detect *SLC45A2* and *G6PC3* Mutations in a Single Patient with Oculocutaneous Albinism and Neutropenia

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Supplementary Tables

Supplementary Table 1. White blood cell counts in the patient on and off Neupogen® treatment.

	On therapy	2 days off therapy	Normal Range
Total WBC count (K/μl)	5.48	1.71	3.4-9.6
Absolute neutrophil count (K/μl)	4.379	0.58	1.29-7.5
Neutrophil (%)	79.9	34	38-78
Absolute lymphocyte count (K/μl)	0.822	0.975	0.48-4.9
Absolute monocyte count (K/μl)	0.27	0.54	0.0-1.5
Absolute eosinophil count (K/μl)	0.00	0.00	0.-0.77
Absolute basophil count (K/μl)	0.0	0.00	0..29

* WBC – White Blood Cell

** K/μl – Thousands of cells per microliter

Supplementary Table 2. Whole blood and immunological analysis

	Patient Values	Normal Range
ALT (U/L)	19	6-41
Complement C3 (mg/dL)	154	69-175
Complement C4 (mg/dL)	24	13-38
Hematocrit (%)	36	31.8-43.2
Hemoglobin (mg/dL)	12.9	11.1-15.0
IgG subclasses 1-4	normal	-
Lymphocyte B-cell defects	normal	-
Lymphocyte phenotyping TBNK	normal	-
Platelets (K/uL)	21	162-380
Prothrombin time (s)	13.3	11.8-14.7
Reticulocyte (%)	1.49	0.66-2.47
Reticulocyte count (K/uL)	60.0	31.7-104.6
Serum creatinine (mg/dL)	0.8	0.7-1.3
Serum sodium (mmol/L)	139	135-144
Total bilirubin (mg/dL)	0.6	0.1-1.0
Total IgA (mg/dL)	88	91-499
Total IgG (mg/dL)	1170	642-1730
Total IgM (mg/dL)	104	34-342

* K/μl – Thousands of cells per microliter

Supplementary Table 3. Filtered gene variants within known homozygous regions of the patient.

	Genome Position				Allele			Amino Acid			CDPred
Chromosome	Left	Right	Gene	Mutation Type	Reference	Variant	Patient	Reference	Variant	Position	Score
5	33990267	33990269	SLC45A2	Frameshift (Stop)	A	-	--	A	PfsX68	330	-30
17	39508724	39508726	G6PC3	Stop	C	T	TT	Q	X	152	-30
17	42590581	42590583	CDC27	Stop	A	C	CC	L	X	155	-30
5	35945878	35945880	CAPSL	Non-synonymous	T	C	CC	D	G	124	-11
19	10684849	10684851	QTRT1	Non-synonymous	C	G	CC	F	L	372	-9
17	42569603	42569605	CDC27	Non-synonymous	T	C	CC	H	R	608	-8
4	42287615	42287617	ATP8A1	Non-synonymous	G	A	AA	L	F	165	-6
17	42569598	42569600	CDC27	Non-synonymous	A	G	GG	F	L	610	-5
17	42574281	42574283	CDC27	Non-synonymous	T	C	CC	Y	C	495	-4
17	42569590	42569592	CDC27	Non-synonymous	T	G	GG	L	F	612	-3
17	42574327	42574329	CDC27	Non-synonymous	A	C	CC	C	G	480	-3
17	42590633	42590635	CDC27	Non-synonymous	A	C	CC	Y	D	138	-3
19	11839827	11839829	ZNF439	Non-synonymous	G	T	TT	R	I	315	-3
19	12724428	12724430	BEST2	Non-synonymous	G	A	AA	R	Q	8	-2
17	42571160	42571162	CDC27	Non-synonymous	A	C	CC	D	E	548	-1
17	42574311	42574313	CDC27	Non-synonymous	T	C	CC	N	S	485	-1
17	42587149	42587151	CDC27	Non-synonymous	A	C	CC	F	V	282	-1
17	42590596	42590598	CDC27	Non-synonymous	G	C	CC	S	C	150	-1
17	42571167	42571169	CDC27	Non-synonymous	T	G	GG	Q	P	546	-1
17	42590651	42590653	CDC27	Non-synonymous	C	T	TT	A	T	132	0
17	42571170	42571172	CDC27	Non-synonymous	A	G	GG	L	P	545	1
17	42576284	42576286	CDC27	Non-synonymous	C	T	TT	S	N	378	1
17	42576316	42576318	CDC27	Non-synonymous	A	C	CC	I	M	367	1
17	45807774	45807776	EME1	Non-synonymous	A	C	CC	E	D	69	2
19	9952350	9952352	COL5A3	Non-synonymous	A	T	TT	D	E	859	2
4	52637817	52637819	SPATA18	Non-synonymous	A	G	GG	K	R	292	2
4	74672483	74672485	RASSF6	Non-synonymous	A	G	GG	Y	H	107	2
17	42574294	42574296	CDC27	Non-synonymous	G	C	CC	P	A	491	3
4	70190004	70190006	UGT2B28	Non-synonymous	G	A	AA	A	T	346	3

Supplementary Table 3 Legend

The whole-exome data were aligned to USCS human genome build 18, and filtered for known SNPs. Data are sorted by the potential detriment (CD_Pred) score. Note the two mutations in *SLC45A2* (c.987delA, p.A330PfsX68) and *G6PC3* (c.829C>T, p.Q277X).

Supplementary Figure Legends

Supplementary Figure 1. Homozygous regions in the patient

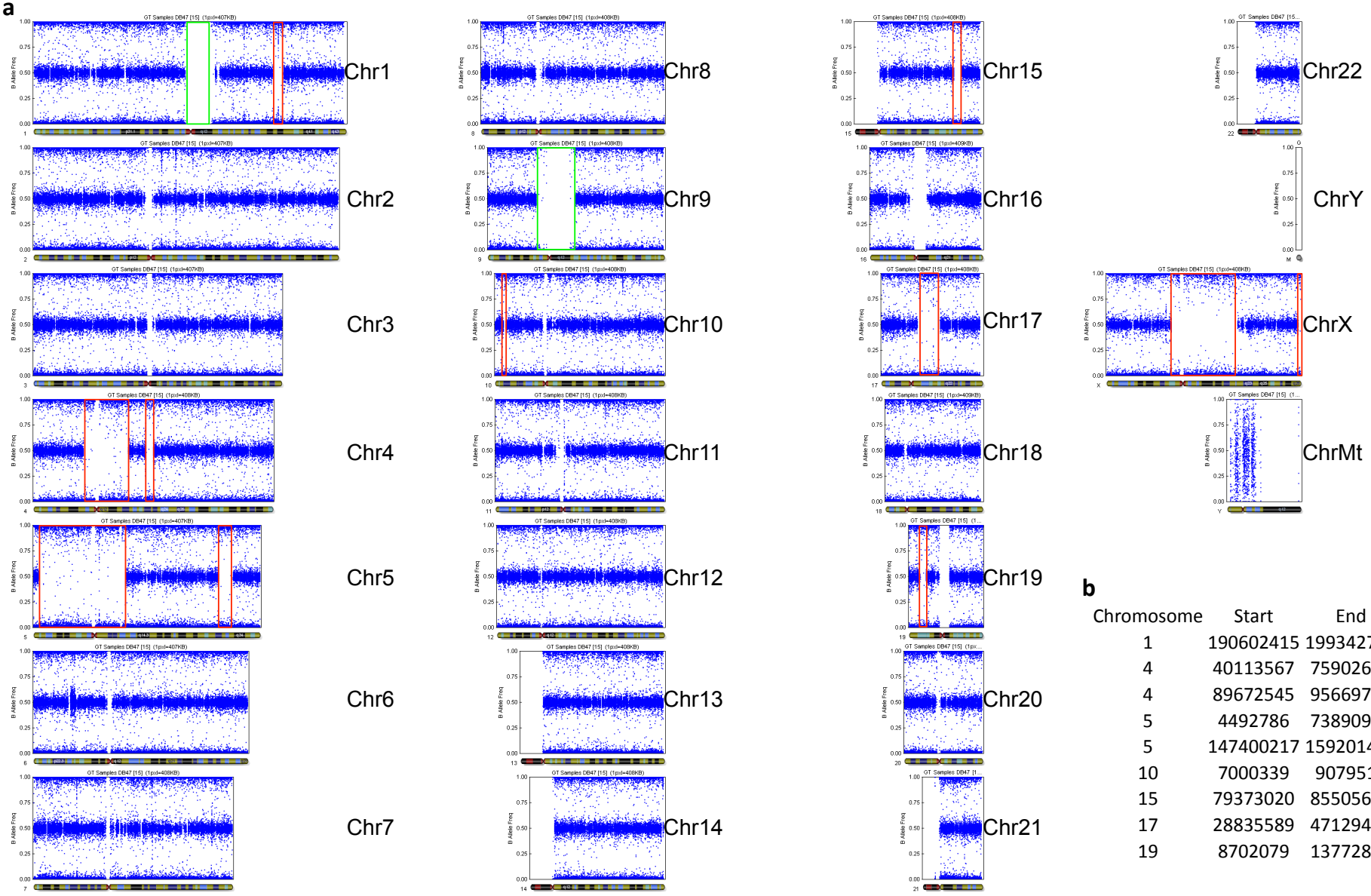
a) SNP data for the patient showing the regions of extended homozygosity (red boxes) on several chromosomes. Normal regions of no-called SNPs are outlined on chromosome 1 and 9 with a green box. The middle of the B allele plots represents heterozygosity (AB) and the upper and lower edges represent homozygosity (AA or BB).
b) Table showing actual start and end positions of extended regions of homozygosity. The base positions were determined from USCS genome build 18.

Supplementary Figure 2. Coomassie and silver stained gels

a) Coomassie stained polyacrylamide gel of conditioned medium and lysates from control and patient cells. The same pattern and intensity of bands can be seen for both control and patient lanes.
b) Silver stained gel from a, showing same pattern and intensity of bands for both control and patient's condition media.

Supplementary Figure 1

a



b

Chromosome	Start	End
1	190602415	199342790
4	40113567	75902618
4	89672545	95669793
5	4492786	73890920
5	147400217	159201457
10	7000339	9079512
15	79373020	85505666
17	28835589	47129438
19	8702079	13772872

Supplementary Figure 2