

Supplementary Information For:

Exome Sequencing Identifies *GATA1* Mutations Resulting in Diamond-Blackfan Anemia

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Supplementary Table 1: Fetal Hemoglobin and Erythrocyte Adenosine Deaminase (eADA) Testing

Individual	eADA Level (EU/grams of Hb)	HbF (% of total hemoglobin)
II-1 (at age 5 years)	0.72	8.4
I-1	0.53	Not done
I-2	0.49	Not done
Notes	Normal range is 0.33 - 0.96	Normal range is < 1 %

Supplementary Table 2: Details of Exome Sequencing in Two Siblings with DBA

Sample	Individual II-1	Individual II-3
No. of bait DNA bases	44,870,454	44,870,454
No. of target DNA bases	32,950,014	32,950,014
Total Reads	124,859,508	134,892,718
Purity Filtered (PF) Unique Reads	82,422,497	85,541,405
PF Unique Reads Aligned	75,799,490	78,493,355
On Target Bases	3,035,025,577	3,167,638,694
Fold Enrichment	51.39	51.54
Target Bases 2x %	97.33	97.32
Target Bases 10x %	93.13	93.58
Target Bases 30x %	83.05	84.31
Total SNPs	19,528	19,607
Percentage in dbSNP	96.71	96.57
No. of SNPs in dbSNP	18,886	18,934
Identified Variants After Filtering Variants in dbSNP Build 132, 1000 Genomes Project, and 95 Exomes from NIEHS Environmental Genome Project		
Total Number	287	289
Silent	99	90
Missense	188	197
Nonsense	0	2
Readthrough	0	0

**Supplementary Table 3: Summary of Genotyping in the DBA Family
(Variants are highlighted in yellow and genotyping failures are highlighted in red)**

Gene	Ref #	Variant pos.(hg19 coordinates)	REF	ALT	I-1	I-2	II-1	II-2	II-3
PLB1	NM_001170585	28843138	G	C	G/C	G/G	G/C	G/C	G/C
TTC27	NM_001193509	33012179	G	A	G/A	G/G	G/A	G/A	G/A
NEB	NM_001164507	152432756	T	C	T/T	T/C	T/C	T/C	T/C
ACVR1C	NM_001111031	158390548	C	T	C/T	C/T	C/T	C/T	C/T
POC1A	NM_001161580	52179931	C	T	C/C	C/T	C/T	C/T	C/T
CYP21A2	NM_000500	32008783	C	T	G/C	G/G	G/C	G/G	G/C
SYNE1	NM_033071	152652136	C	T	C/T	C/T	C/T	C/T	C/T
PDLIM2	NM_0241630	22447111	T	C	T/C	T/T	T/C	T/C	T/C
FAM129B	NM_001035534	130272445	C	T	C/T	C/C	C/T	C/C	C/T
CDHR1	NM_001171971	85961593	C	T	C/C	C/T	C/T	C/T	C/T
C10orf125	NM_001098483	135168915	T	G	T/T	T/G	T/G	T/G	T/G
ART5	NM_001079536	3660256	C	A	C/A	C/C	C/A	C/A	C/A
CACNA1H	NM_001005407	1254048	G	T	G/T	G/G	G/T	G/T	G/T
PDPK1	NM_002613	2607963	C	T	C/C	C/T	C/T	C/T	C/T
MVP	NM_005115	29859222	G	T	G/T	G/G	G/T	G/G	G/T
CNTR0B	NM_001037144	7852817	G	A	G/A	G/G	G/A	G/G	G/A
NCOR1	NM_001190440	15967434	C	G	C/G	C/C	C/G	C/C	C/G
C17orf70	NM_025161	79514391	C	T	C/T	C/C	C/T	C/C	C/T
SLA2	NM_032214	35262950	G	T	G/G	G/T	G/T	G/G	G/T
RFPL3	NM_001098535	32754252	A	G	variant not present	variant not present	variant not present	variant not present	variant not present
ESPN	NM_031475	6505817	C	T	C/T	C/T	C/T	C/T	C/T
CPT2	NM_000098	53676371	T	C	T/T	T/C	T/C	T/C	T/C
FLG	NM_002016	152276050	C	A	C/C	C/A	C/A	C/A	C/A
FLG	NM_002016	152276545	G	A	G/G	G/A	G/A	G/A	G/A
FCRL2	NM_030764	157736647	G	A	G/G	G/A	G/A	G/A	G/A
F13B	NM_001994	197026161	G	C	G/C	G/G	G/C	G/G	G/C
PLEKHH2	NM_172069	43922319	A	G	A/A	A/G	A/G	A/A	A/G
DOCK10	NM_014689	225710038	C	A	C/C	C/A	C/A	C/A	C/A
ARPP21	NM_016300	35833886	T	C	T/T	T/C	T/C	T/T	T/C
IFRD2	NM_006764	50326254	G	A	G/A	G/G			
DNAH1	NM_015512	52400807	G	T	G/T	G/G	G/T	G/G	G/T
DNAH1	NM_015512	52425264	A	G	A/A	A/G	A/G	A/A	A/G
LEKR1	NM_001004316	156710978	A	G	A/G	A/A	A/G	A/A	A/G
NLGN1	NM_014932	173998740	G	T	G/T	G/G	G/T	G/G	G/T
SLC25A31	NM_031291	128651703	G	T	G/G	G/T	G/T	G/T	G/T
C4orf45	NM_152543	159894418	G	T	G/T	G/G	G/T	G/T	G/T
GALNT7	NM_017423	174090089	C	T	C/T	C/C	C/T	C/T	C/T
SCGB3A2	NM_054023	147261026	A	C	A/C	A/A	A/A	A/C	A/C
NUP153	NM_005124	17616339	G	A	G/A	G/G	G/A	G/G	G/A
HIST1H2BG	NM_003518	26216600	G	A	G/A	G/G	G/A	G/G	G/A
ZSCAN16	NM_025231	28097614	G	C	G/G	G/C	G/C	G/G	G/C
NKAPL	NM_001007531	28227993	G	A	G/G	G/A	G/A	G/G	G/A
TNXB	NM_019105	32038017	T	C	T/C	T/T	T/C	T/T	T/C
TREML2	NM_024807	41162551	T	A	T/A	T/T	T/A	T/T	T/A
KHDRBS2	NM_152688	62407136	C	T	C/T	C/C	C/T	C/C	C/T
TRDN	NM_006073	123673686	T	C	T/T	T/C	T/C	T/T	T/C
WDR27	NM_182552	170043831	A	C	A/A	A/C	A/C	A/C	A/C
SMO	NM_005631	128852162	A	G	A/A	A/G	A/G	A/G	A/G
RIMS2	NM_001100117	104778513	C	G	C/G	C/C	C/G	C/G	C/G
KIAA0196	NM_014846	126068971	C	T	C/C	C/T	C/T	C/C	C/T
MAPK15	NM_139021	144801223	C	T	C/T	C/C	C/T	C/T	C/T
TAF1L	NM_153809	32633036	C	T	C/C	C/T	C/T	C/T	C/T
TMC1	NM_138691	75445414	C	T	C/C	C/T	C/T	C/T	C/T
NFIL3	NM_005384	94172680	C	T	C/T	C/C	C/T	C/C	C/T
SLC2A8	NM_014580	130160385	G	T	G/G	G/T	G/T	G/T	G/T
OR5P2	NM_153444	7817570	G	C	G/C	G/G	G/C	G/C	G/C
OR8K3	NM_001005202	56086498	C	T	C/T	C/C	C/T	C/T	C/T
DBB1	NM_001923	61091561	A	G	A/A	A/G	A/G	A/G	A/G
LRP1	NM_002332	57599365	G	C	G/C	G/G	G/C	G/C	G/C
CEP290	NM_025114	88483047	C	T	C/C	C/T	C/T	C/C	C/T
WDFY2	NM_052950	52234780	C	G	C/G	C/C	C/G	C/G	C/G
ABCC4	NM_005845	95714964	T	C	T/T	T/C	T/C	T/C	T/C
ZIC5	NM_033132	100617644	C	T	C/C	C/T	C/T	C/T	C/T
SLC10A2	NM_000452	103718370	C	T	C/C	C/T	C/T	C/T	C/T
SPTBN5	NM_016642	42147913	G	A	G/A	G/G	G/A	G/G	G/A
PLA2G4E	NM_001080490	42279511	C	T	C/T	C/C	C/T	C/C	C/T
ZNF609	NM_015042	64970626	C	A	C/C	C/A	C/A	C/A	C/A
FAM192A	NM_024946	57188379	A	T	A/T	A/A	A/T	A/A	A/T
RI TPR	NM_001013838	67691414	A	G	A/A	A/G	A/G	A/A	A/G
SRP68	NM_014230	74056418	T	C	T/T	T/C		T/T	T/C
FASN	NM_004104	80049154	C	A	C/A	C/C	C/A	C/C	C/A
ADAMTS10	NM_030957	8661029	G	A	G/A	G/G	G/A	G/A	G/A
P2RX1	NM_002558	3808223	C	T	C/T	C/T	C/T	C/T	T

Supplementary Table 4: Hematologic Parameters for DBA Patient with *GATA1* chrX: 48,649,736 Deletion

Age (years, months)	6 weeks old	3.5 years old
WBC x 10⁹/L	6.8	9.3
ANC X 10⁹/L	1.564	2.697
HGB g/dL	3.5	10.9
Platelet x 10⁹/L	362,000	250,000
Fetal Hemoglobin %	83.9	Not done
Erythrocyte Adenosine deaminase EU/g hemoglobin	0.81	Not done
Notes	Time of first presentation	Receiving corticosteroids for over 2 years