

Additional File 1 Supplementary Tables

Table S1: Clinical genetic testing summary

Test	Description	Disease Association	Chr. Location (hg19)	Result: nucleotide change	Result: amino acid change	Frequency (ExAC)	Classification
<i>NPHS2</i> (NM_014625) sequencing	Nephrosis 2, idiopathic, steroid-resistant (podocin)	Nephrotic syndrome/glomerulosclerosis (MIM: 600995)	NA	No mutation detected	NA	NA	NA
<i>LAMB2</i> (NM_002292) sequencing	Laminin, beta 2 (laminin S)	Nephrotic syndrome (MIM: 614199)	NA	No mutation detected	NA	NA	NA
<i>WT1</i> (NM_024426) sequencing	Wilms tumor 1	Nephrotic syndrome (MIM: 256370)	11:32417962	c.1099-9T>C	NA	0.004435	Benign
<i>NPHS1</i> (NM_004646) sequencing	Nephrosis 1, congenital, Finnish type (nephrin)	Nephrotic syndrome (MIM: 256300)	19:36339295	c.1175 T>C	p.Leu392Pro	0.008783	Benign
<i>ACTN4</i> (NM_004924) sequencing	Actinin, alpha 4	Glomerulosclerosis (MIM: 603278)	NA	No mutation detected	NA	NA	NA
<i>TRPC6</i> (NM_004621) sequencing	Transient receptor potential cation channel, subfamily C, member 6	Glomerulosclerosis (MIM: 603965)	NA	No mutation detected	NA	NA	NA
<i>GLIS2/NPHP7</i> (NM_001318918) sequencing	GLIS family zinc finger 2	Nephronophthisis (MIM: 611498)	16:4386814	c.864T>C	p.Tyr288Tyr	0.9796	Benign
<i>IQCB1/NPHP5</i> (NM_001023570) sequencing	IQ motif containing B1	Senior-Loken syndrome (MIM: 609254)	3:121547527	c.101-48 T>C	NA	Not listed	Intronic
			3:121526204	c.574C>T	p.Leu192Leu	0.6275	Benign
<i>NPHP3</i> (NM_153240) sequencing	Nephronophthisis 3 (adolescent)	Nephronophthisis (MIM: 604387)	NA	No mutation detected	NA	NA	NA
<i>SDCCAG8</i> (NM_006642) sequencing	Serologically defined colon cancer antigen 8	Senior-Loken syndrome (MIM: 613615)	1:243507680	c.1473+48dupA	NA	0.2608	Intronic
			1:243579112	c.1725G>A	p.Glu575Glu	0.4889	Benign
			1:243493907	c.1134A>T	p.Glu378Asp	0.2976	Benign
<i>INVS/NPHP2</i> (NM_014425) sequencing	Inversin	Nephronophthisis (MIM: 602088)	9:103059476	c.3016+48 A>G	NA	0.6803	Intronic
Chromosome microarray (Affymetrix 6.0, 1.8 Mb oligo-SNP)		NA	16p13.3p12.3 (hg19: chr16:94807-20250946) x2 hmz	NA	NA	NA	NA

NA, Not applicable

Table S2. Whole exome sequencing capture statistics

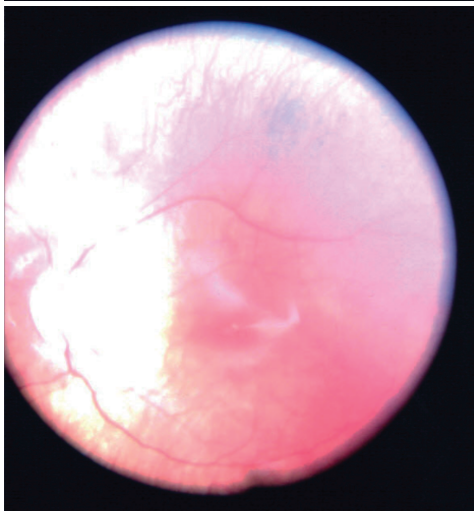
Sample Name	Sample Role	Total Pass Filter (Gb)	% Duplicate Reads	% Total Reads Aligned	Avg Coverage	Reads Hit Target/ Buffer	% Targets Hit	% Base 1+ Coverage	% Base 20+ Coverage	% Base 40+ Coverage
DM165-1000	Father	9	3%	99%	102	77%	99%	99.01%	95.88%	86.75%
DM165-1001	Mother	12	4%	99%	146	77%	99%	99.05%	96.92%	93.28%
DM165-0001	Affected	9	3%	99%	103	77%	99%	99.03%	95.89%	86.66%

Table S3. Final variant list following trio-based exome analysis (<1% minor allele frequency (MAF), homozygous, compound heterozygous, de novo, or X-linked).

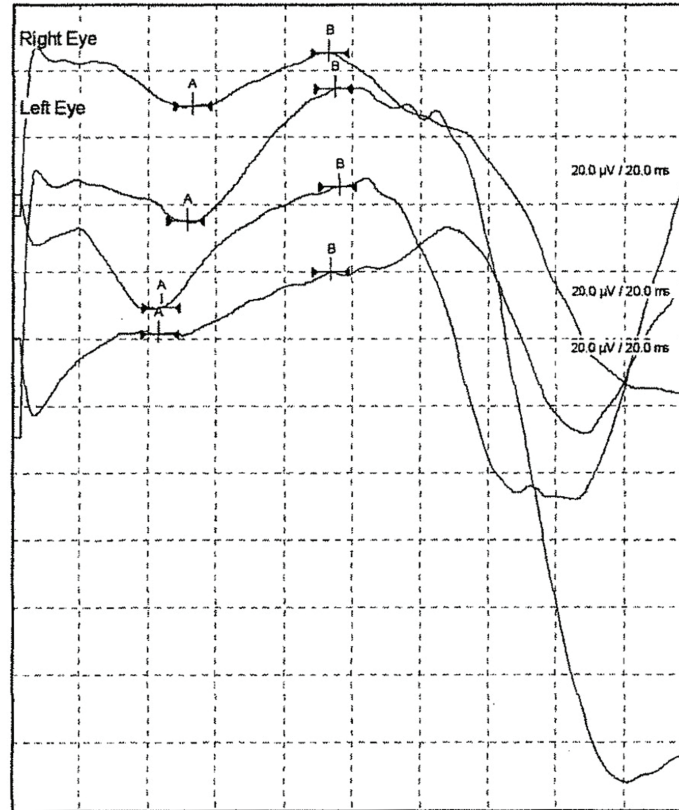
Gene	Description	Disease Association	Chr:Location (hg19); rsID	Nucleotide	Amino acid	Zygosity	Inheritance	Total ExAC freq
<i>IFT140</i>	Intraflagellar transport 140 homolog (Chlamydomonas)	Mainzer-Saldino Syndrome (MIM: 266920)	16:1642177 (rs201188361)	c.634G>A exon 6 donor splice site	p.G212R	UPD Hom	Mat	4.121e-05
<i>ABCA3</i>	ATP-binding cassette, sub-family A (ABC1), member 3	Surfactant metabolism dysfunction, pulmonary, 3 (MIM: 610921)	16:2329071 (rs146709251)	c.4420C>T	p.R1474W	UPD Hom	Mat	0.005275*
<i>ABCC1</i>	ATP-binding cassette, sub-family C (CFTR/MRP), member 1		16:16110432	c.569G>A	p.C190Y	UPD Hom	Mat	0
<i>ARHGEF10</i>	Rho guanine nucleotide exchange factor (GEF) 10	Slowed nerve conduction velocity, AD (MIM: 608236)	8:1846642	c.1601_1603 delTGA	p.M535del	Het	AR	0
<i>ARHGEF10</i>	Rho guanine nucleotide exchange factor (GEF) 10	Slowed nerve conduction velocity, AD (MIM: 608236)	8:1905361 (rs201516531)	c.3967C>T	p.R1323W	Het	AR	7.267e-05

* ≥ 1 Homozygotes present in ExAC (accessed March 2017). Abbreviations: ExAC, Exome Aggregation Consortium; UPD, uniparental isodisomy; Mat, maternal; Pat, paternal; AR, autosomal recessive; MIM, Mendelian inheritance in man.

a



b



c

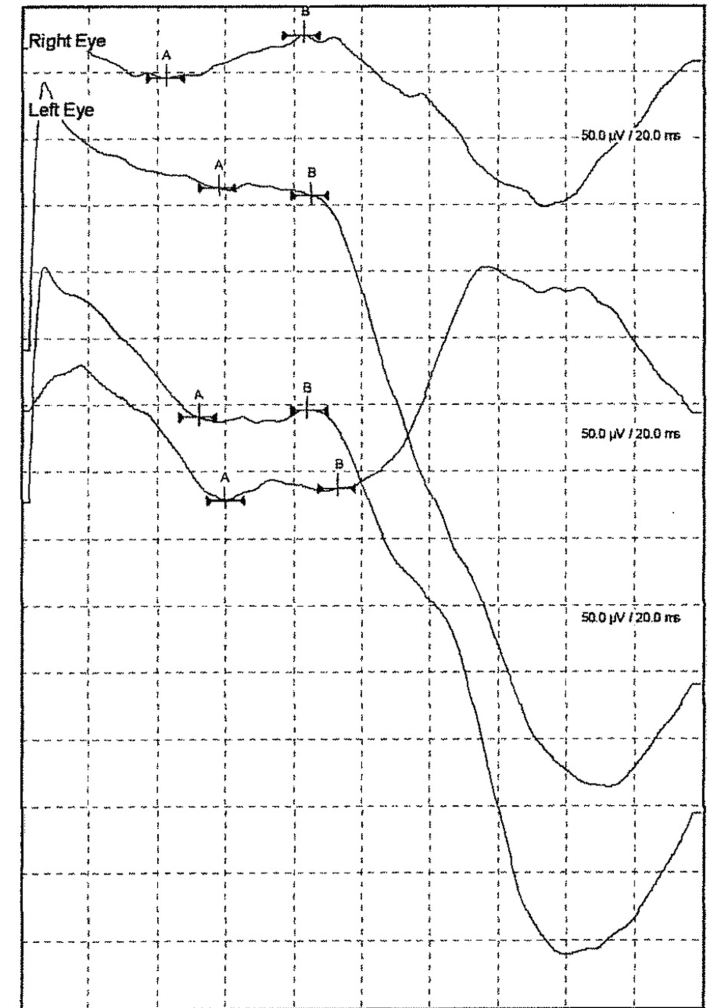


Figure S1

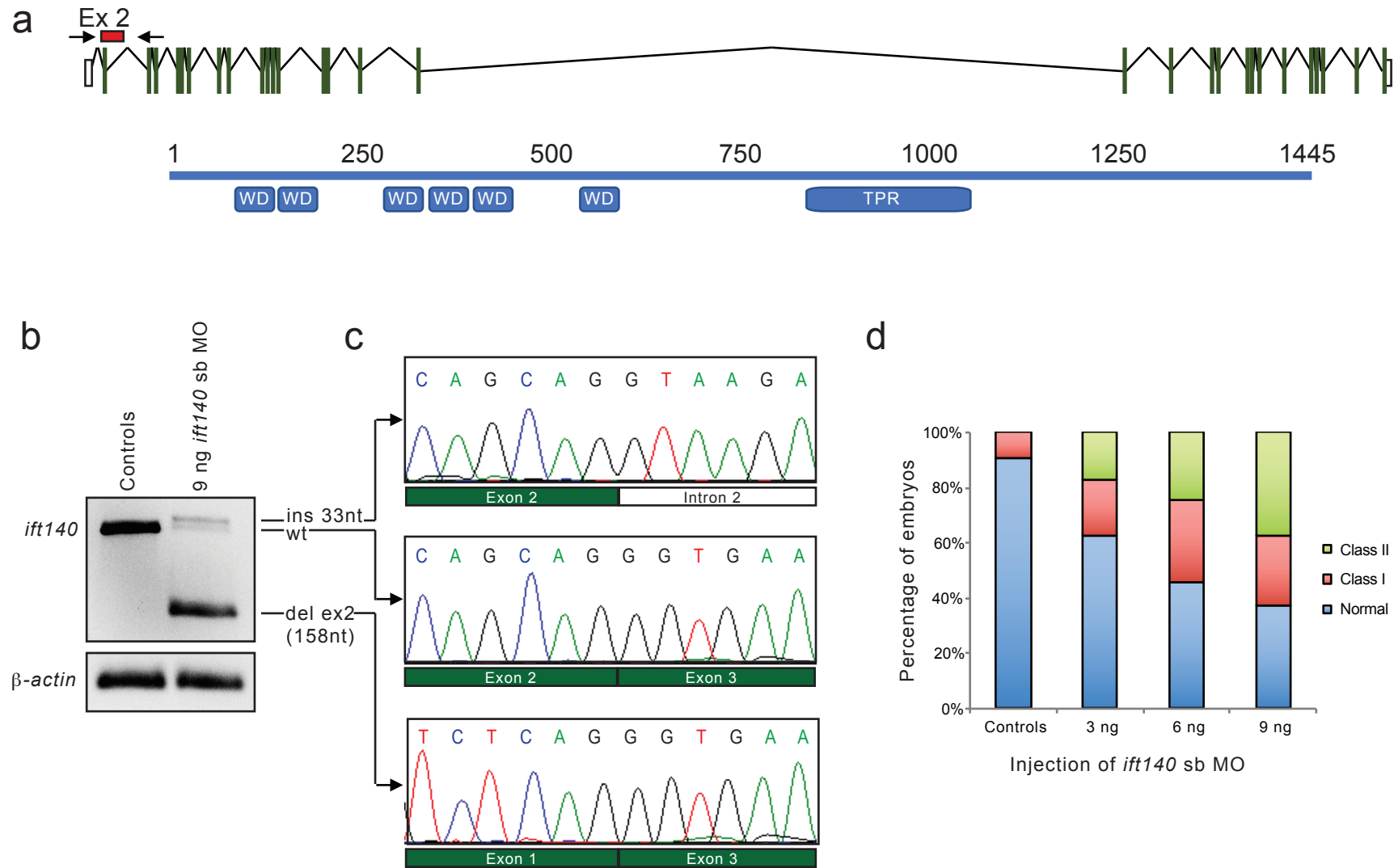


Figure S2

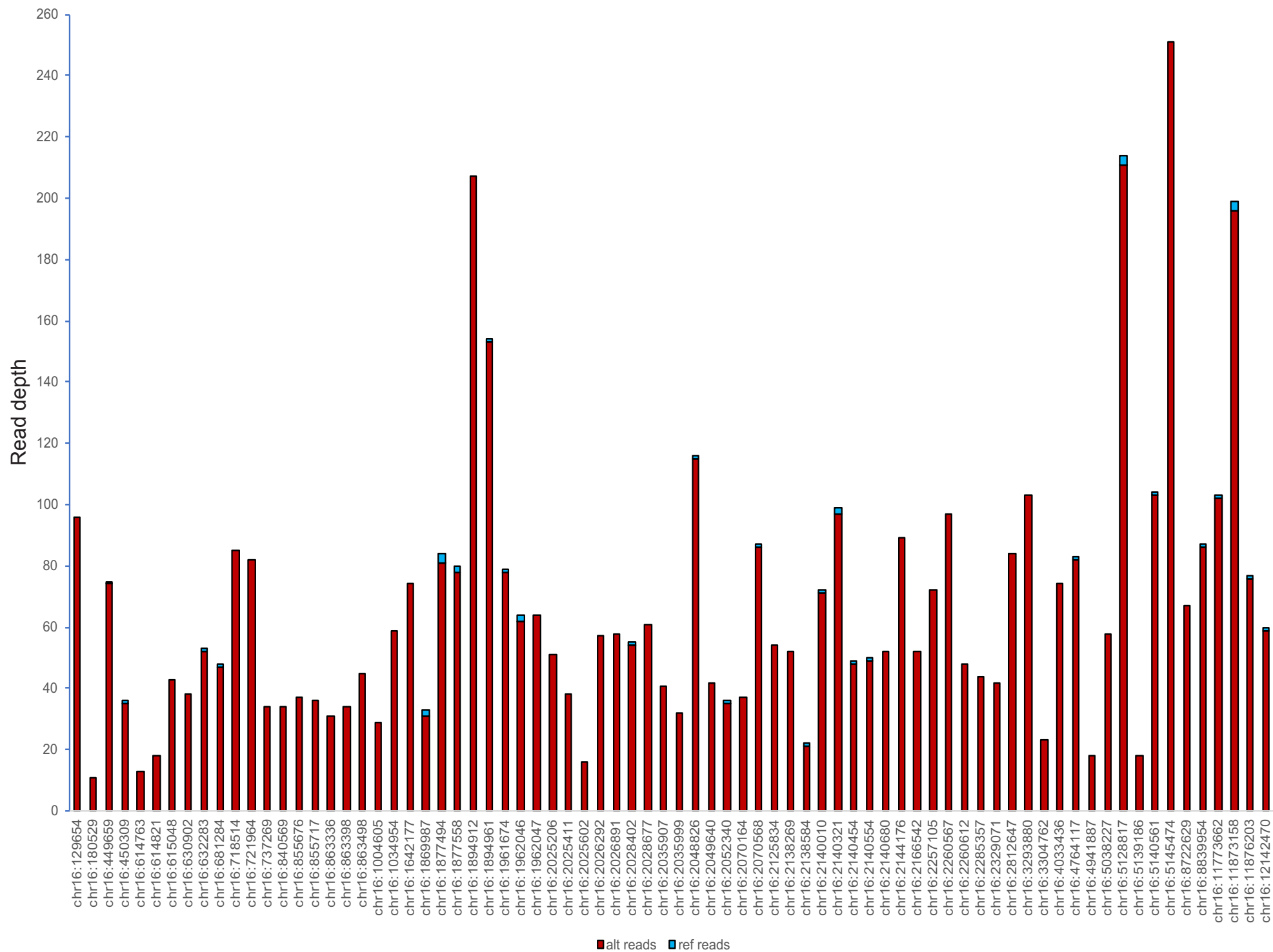


Figure S3