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Supplemental Data

Exome Sequencing Reveals De Novo *WDR45* Mutations Causing a Phenotypically Distinct, X-Linked Dominant Form of NBIA

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Supplemental Table 1. Sequencing Statistics

				Mapped	Reads	Target Bases			
Patient ID	Reads	Mapped Reads	Percent	Sequence (Gb)	Overlapping Target	>1x	>8x	>20x	Average Coverage
#60251	97,206,974	93,571,072	96.26	9.82	69.83	99.64	97.47	93.16	103.31
#63700	83,978,185	82,704,872	98.48	8.48	78.95	99.54	96.77	91.74	105.02
#63701	104,714,028	102,878,906	98.25	10.58	77.4	99.53	96.90	92.58	126.86
#63702	97,994,022	96,369,023	98.34	9.90	77.73	99.54	97.02	92.67	119.39
#63703	98,730,027	97,062,160	98.31	9.97	78.47	99.58	97.35	93.10	121.94
#63704	103,855,311	102,184,278	98.39	10.49	80.12	99.60	97.48	93.64	131.38
#63705	105,151,908	103,338,107	98.28	10.62	78.61	99.60	97.35	93.36	130.8
#63706	119,774,913	118,099,728	98.6	12.10	76.50	99.64	97.57	93.89	144.49
#63707	103,127,329	101,520,019	98.44	10.42	77.61	99.61	97.53	93.67	126.41
#63708	76,819,820	74,617,432	97.13	7.76	79.42	99.90	97.95	92.16	94.97
#63709	103,233,138	101,703,042	98.52	10.43	77.95	99.57	97.19	93.07	126.85
#63710	99,932,444	98,094,726	98.16	10.09	77.31	99.60	96.76	90.40	121.31
#63711	106,227,473	104,578,095	98.45	10.73	77.33	99.52	96.89	92.68	128.85
#63712	111,916,818	109,971,381	98.26	11.3	77.97	99.60	97.53	93.85	137.37
Average	**100,904,456**	**99,049,489**	**98.13**	**10.19**	**77.51**	**99.61**	**97.27**	**92.86**	**122.78**

Supplemental Table 2. Variants Identified in 14 Individuals with BPAN by Exome Sequencing

Patient ID	#60251	#63700	#63701	#63702	#63703	#63704	#63705	#63706	#63707	#63708	#63709	#63710	#63711	#63712
synonymous	10,391	10,221	10,411	12,574	12,422	11,160	10,602	10,616	10,523	10,476	10,553	10,369	10,331	10,480
NSV	11,332	11,332	11,465	14,082	13,898	12,273	11,758	11,604	11,532	11,514	11,482	11,435	11,518	11,408
rare NSV	294	274	234	2,116	1,866	753	312	245	301	291	252	240	260	224
genes with ≥2 NSV	6	13	8	392	345	70	10	6	17	12	10	7	10	7

Non-synonymous variants (NSV) = missense, nonsense, stoploss, splice site disruption, insertions, deletions; rare indicates a frequency <0.3% in 1429 control exomes, HapMap, and the 1000 Genomes project; bold indicates gene carrying the causal mutations.