Prioritization of neurodevelopmental disease genes by discovery of new mutations

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Supplementary Table 1 Genes implicated in developmental disorders (germline mutations) and cancer (somatic mutations)

Gene	Germline mutations			Somatic mutations		
	Neurodevelopmental disorder	PubMed ID	Mutation types	Cancer types / malignancies	PubMed ID	Mutation types
Isolated neurodevelopmental phenotypes and cancer						
PTEN	ASD, Cowden syndrome[a]	23160955, 9259288	LoF, missense	Many types	21252315	Missense, LoF
CTCF	ID	23746550	LoF	Breast cancer, prostate cancer	9591631	Deletion
ARID1B (SWI/SNF complex)	ID, ASD, Coffin-Siris syndrome	22426309, 22426308	LoF	Many types	22426308	LoF
Clinically defined neurodevelopmental syndromes and cancer						
MED12	Lujan-Fryns syndrome, Ohdo syndrome, Opitz-Kaveggia syndrome	17369503, 23395478,	Missense	Prostate cancer	22610119	Missense
MLL2 (*KMT2D*)	Kabuki syndrome	20711175	LoF, CNV, missense	Many types	21796119, 21163964	LoF, missense
CREBBP	Rubinstein-Taybi syndrome; epilepsy	7630403, 11331617	LoF, CNV, missense	ALL	21390130	LoF, missense
ATRX	X-linked α-thalassemia–mental retardation syndrome	7697714	Missense, LoF	PanNET, glioblastoma	21252315, 22286061	Missense, LoF
Identical mutations in neurodevelopmental phenotypes and cancer						
ASXL1	Bohring-Opitz syndrome	21706002	LoF	Myeloid malignancies	19388938	LoF
SETBP1	Schinzel-Giedion syndrome	20436468	GoF	Leukemia	23222956	GoF
EZH2	Weaver syndrome[a]	22177091	GoF	B-cell lymphoma	20081860	GoF
'Paternal age–effect disorders' (*FGFR2*, *FGFR3*, *HRAS*, *PTPN11*, *BRAF*, *MAP2K1*)	Apert syndrome, Crouzon-Pfeiffer syndrome, achondroplasia, Muenke syndrome, Costello syndrome[a], Noonan syndrome[a], cardio-facio-cutaneous syndrome	22325359	GoF	Many types	22325359	GoF
Different mutation types observed in neurodevelopmental phenotypes and cancer						
CTNNB1	ID and ASD	23033978	LoF	Many types	12060769	Activating?, LoF?
CHD7	CHARGE syndrome	15300250	CNVs, LoF, missense	Small-cell lung cancer	20016488	Duplications, CNV, amplifications, translocations
MYCN	Feingold syndrome	15821734	LoF, missense	Neuroblastoma	6197179	Amplification
ABCC9	Cantu syndrome	22610116, 22608503	GoF	Endometrial cancer	23104009	Missense

PanNET, pancreatic neuroendocrine tumors; ALL, acute lymphoblastic leukemia; AML, acute myeloid leukemia.
[a]Patients have increased risk of cancer.