Table 1 *A molecular classification of congenital/developmental hydrocephalus.*

Disease	Broad etiologic category	Specific genetic associations
X-linked hydrocephalus with congenital aqueduct stenosis	X-linked hydrocephalus	L1-CAM
CRASH syndrome		
Fried-type syndrome		AP1S2
Myelomeningocele	Neural tube defects	Genes related to neural tube development, ciliary beating
		Genes related to the folate-homocysteine metabolic pathways
Dandy-Walker malformation	Dandy-Walker malformation	Chromosomal abnormalities in 2q, 5p, 8p, 9p, 13q, 16q, 17q
		Gene mutations: POMT1, POMT2, POMGNT1, FKRP, FKTN, ISPD, LARGE
Holoprosencephaly	Holoprosencephaly	Trisomy 13 (most common) and trisomy 18
		Mutations in 7-dehydrocholesterol reductase as well as SHH, ZIC2, SIX3, TGIF etc.
Primary Ciliary Dyskinesia	Ciliopathies	Genes related to ciliary structure and function, e.g. DNAH11, NEK10 and GAS2L2
Other motile ciliopathies		CCNO and MCIDAS mutations
Nonsyndromic autosomal recessive hydrocephalus	Nonsyndromic autosomal recessive hydrocephalus	Variations of MPDZ and CCDC88C genes
Joubert syndrome and Meckel syndrome	Hydrocephalus plus obstructive arachnoid cyst	CC2D2A gene mutations
Phelan-McDermid syndrome		22q13.3 deletion
Noonan syndrome	RAS-opathies	Mutations in RAS pathway (e.g. NF1, BRAF, KRAS, PTPN11)
Cardio-facio-cutaneous syndrome		
Costello syndrome		
Megalencephaly syndromes	Megalencephaly syndromes	Mutations in genes involved in the PI3K-AKT pathway
Craniosynostosis syndromes	Craniosynostosis syndromes	Mutations in fibroblast growth factor receptor (FGFR) genes
VACTERL-H sequence	VACTERL-H sequence	Mutations in the FANCB gene