Supplementary Table 1

Clinical details of Cohort 3, with 45 individuals with congenital microcephaly and additional features

Individual	Features additional to microcepha	Sex	Ethnicity	*ASPM* mutation
1		F	Caucasian	
2	JEJ	M	Caucasian	
3	ACC, SZ	F	Caucasian	
4	AMC	F	Caucasian	
5	Partial ACC, XAX	F	Caucasian	
6	ACC	F	Caucasian	
7	XAX, BS-CBLH	M	Caucasian	
8	Cervical cord astrocytoma	M	Caucasian	
9	XAX, SQ, SZ	F	Caucasian	
10	ACC, CVH	F	Caucasian	
11	Partial ACC, CVH, SQ	M	Asian	
12	XAX, BS-CBLH, SQ, SZ	F	Caucasian	
13		M	Asian	
14		M	Asian	c.3811C>T
15	ACC, XAX, JEJ	M	Caucasian	
16	EYE/AntCh, JEJ	F	Caucasian	
17	SZ, vocal cord paralysis	M	Caucasian	
18	CVH	M	Caucasian	
19		M	Caucasian	
20		M	Caucasian	
21	CVH, XAX, SZ	F	Caucasian	
22	BS-CBLH	F	Caucasian	
23	SZ	M	Caucasian	
24	BS-CBLH	M	Caucasian	
25		M	Caucasian	
26	BS-CBLH	M	Caucasian	
27		M	Caucasian	c.9178C>T
28	XAX, lymphedema, RET	F	Caucasian	
29	ACC, BS-CBLH, XAX	M	Caucasian	
30	BS-CBLH, XAX	M	Caucasian	
31	Partial ACC	M	Caucasian	
32	XAX	M	Caucasian	
33		F	Caucasian	
34	BS-CBLH (CASK)	M	Caucasian	
35	SQ, SZ, hirsutism, syndromic	U	Caucasian	
36	SQ, SZ	F	Caucasian	
37	SQ, SZ	F	Caucasian	
38		M	Caucasian	
39		M	Caucasian	c.1152_53delAG and c.3710C>G
40	XAX	F	Asian	
41		F	Caucasian	
42		M	Caucasian	
43	ACC, BS-CBLH, SZ	F	Caucasian	
44		M	Caucasian	
45	XAX, SZ	M	Black	

Key to additonal features:

ACC, agenesis of the corpus callosum
AMC, arthrogryposis multiplex congenita
BS/CBLH, disproportionate brainstem and cerebellar hypoplasia
CVH, cerebellar vermis hypoplasia
EYE/AntCh, anterior chamber anomalies
JEJ, jejunal atresia
RET, retinal abnormality
SQ, spastic quadriparesis
SZ, seizures
XAX, enlarged extra-axial space