

Appendix I. DNA isolation according to the Manual of Center of Biomedical Research (Faradz, 2004)

Salt saturation method was performed. EDTA frozen blood was transferred into a 50 ml tube with the addition of NH₄Cl 5-10 ml lysis buffer. The tube was incubated for 10–30 minutes at room temperature. Afterwards the tube was centrifuged for 5 minutes at 3000-3500 RPM, followed by removal of the supernatant and addition of NH₄Cl lysis buffer. The procedure was repeated three times. TE lysis buffer 2 ul, Proteinase-K 10mg/ml and 100 ul 10% SDS were added and mixed until white pellet was formed. The tube was then incubated at 50°C for 24 hours. After incubation NaCl 6M approximately one third volume of the tube was added to the suspension and centrifuged at 4000 RPM for 10 minutes. The supernatant was moved to a new tube and absolute ethanol twice the volume of the supernatant was added. The DNA formed white viscous thread and could be removed by fine needle, rinsed with 70% ethanol and moved to a new 1,5 ml tube. The new tube was left open to allow evaporation of ethanol. The tube was then filled with TE buffer for dissolution of the DNA. (Faradz, 2004)

Appendix II. The Nellhaus charts

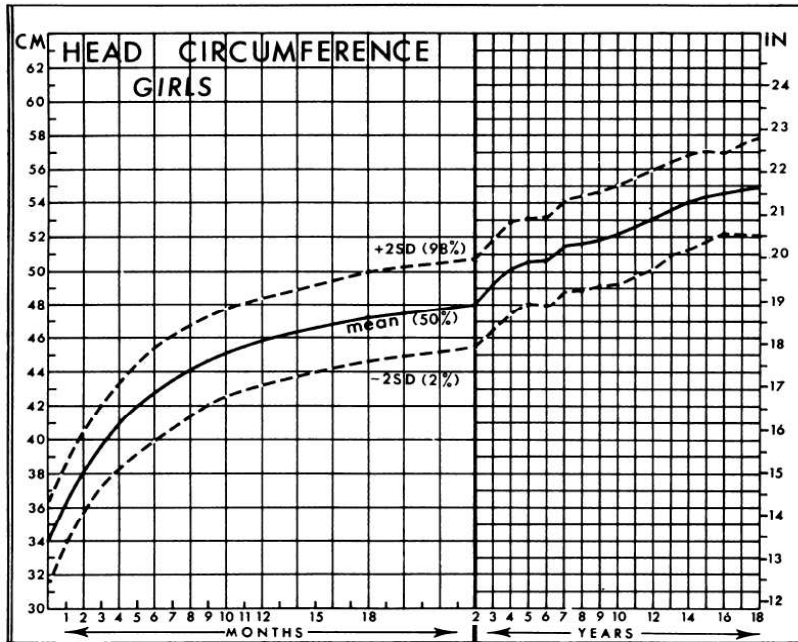
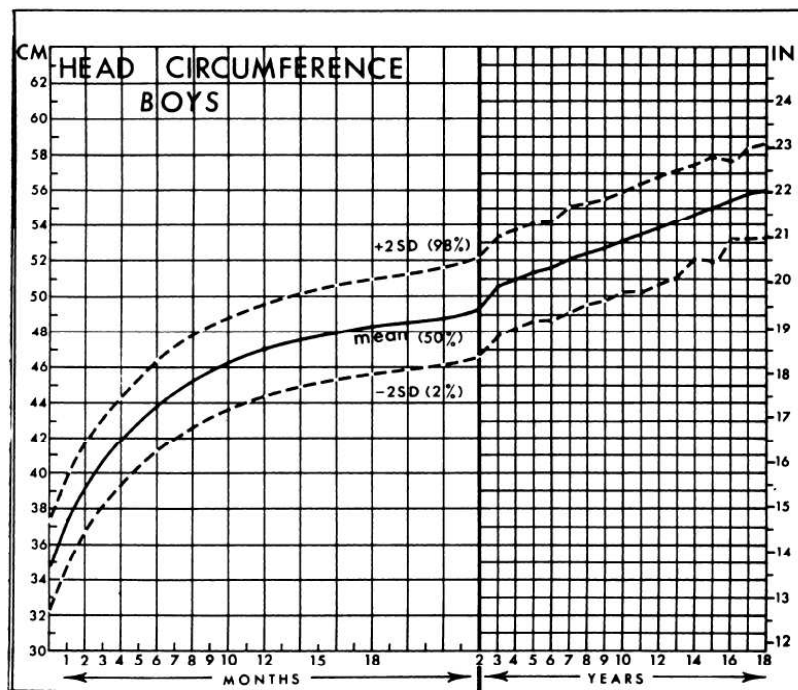


FIG. 2. Composite graph for females from birth through 18 years.



Appendix III. List of primers

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	R	gtgcactgaggaaggaccaa
EXON 17-18	F	agtcccatgtgctgtgcat
	R	tgacaccaagactcccacaa
EXON 19	F	ctgtggtgtgggtggctt
	R	ctgtcttggccaccactgt
EXON 20	F	acatggagcttagcacaggg
	R	tatgtgccaggccctgtaaa
EXON 21	F	gttgccctctttggggactg
	R	agggaaaagagggacatgga
EXON 22	F	agagtccagatgggctgtgt
	R	tggagtgggagtcagcctaa
EXON 23	F	gttcacagcctccagcagt
	R	ggcagagagcacttcttcca

EXON 24	F	gtacaggtgcctaggggctc
	R	gcttcagtactgggtcaaagtca
EXON 25-26	F	taccatccctcctccagatg
	R	ggagccaactccagggac
EXON 27-29	F	gggtttctggggagtgc
	R	acgccccacagctaggc
EXON 30_1	F	gatgtccagcctagctgtgg
	R	cagggtcagtctcaggttg
EXON 30_2	F	gcttcaggccatcaccac
	R	gaatgaatggcacagtccct
EXON 31	F	agtacagcatggagtccacc
	R	gactgagctgggtctccacctt
EXON 32	F	ctaagccccagagttggga
	R	caggaaccattcctcaaagc

Appendix IV. List of Single Nucleotide Polymorphisms

The list of Single Nucleotide Polimorphism (SNP) of *ASPM* gene among children with microcephaly

rs Number	SNP	n	Cohort Total	Present study %	European %
rs1571964	c.441+14C>T	43	48	89.6	95.8
rs6677082	c.849C>T	29	48	60.4	91.2
rs4915337	c.3579T>A	43	48	89.6	6.8
rs964201	c.7480T>C	45	48	93.8	99.1
rs1412640	c.7566A>G	46	48	95.8	91.2
rs10922162	c.7605G>A	42	48	87.5	14.3
rs10754213	c.10331+8A>G	44	48	91.7	91.6
rs4915344	c.2174-20T>C	8	48	16.7	18.8
rs6676084	c.3138G>A	7	48	14.6	38.3
rs2878749	c.4449A>G	10	48	20.8	38.9
rs41310925	c.5961A>G	10	48	20.8	NA
rs41308365	c.7674C>T	9	48	18.8	NA
rs41310927	c.7684A>G	10	48	20.8	NA
rs3762271	c.7939C>A	9	48	18.8	NA
rs12138336	c.7860G>C	1	48	2.1	6.7

The list of Single Nucleotide Polimorphism (SNP) of *WDR62* gene among children with microcephaly

rs Number	SNP	n	Present study total	Present study %	European %
rs61742664	c.180G>A	16	48	30	17.5
rs11538454	c.186C>T	7	48	14.6	20.4
rs78138007	c.562-13C>T	18	48	37.5	1.7
rs76130844	c.1233+13C>T	20	48	41.7	5.8
rs2301734	c.1641G>A	11	48	22.9	21
rs61741470	c.3401T>G	20	48	41.7	5
rs45470992	c.3639C>T	20	48	41.7	5
rs2074435	c.3929A>T	42	48	87.5	60.3
rs1008328	c.4170A>C	10	48	20.8	70.5
rs2285745	c.2549T>C	31	48	64.6	70.4
rs10423651	c.700-18C>T	15	48	31.3	35
rs2301734	c.1641G>A	21	48	43.8	NA
rs118175551	c.3033G>A	5	48	10.4	NA

*NA = not available

The list of Single Nucleotide Polimorphism of *MCPH1* gene among microcephaly children

rs Number	SNP	n	Present study total	Present study %	European %
rs2305022	c.228G>T	9	20	45	77.9
rs2305023	c.-58G>C	9	20	45	22.5
rs2445313	c.513T>A	10	20	50	0
rs930557	c.940G>C	12	20	60	78.3
rs2515569	c.1175A>G	19	20	95	98.6
rs2936531	c.2215-19A>G	18	20	90	46.6
rs2912010	c.2226C>T	16	20	80	44.7
rs1057090	c.2282C>T	16	20	80	39.2
rs1057091	c.2482C>T	4	20	20	73
rs2442513	c.513T>G	5	20	25	38.4
rs2920676	c.1428C>T	1	20	5	2.7
rs75741316	c.647T>C	5	20	25	NA
rs2920676	c.1428C>T	4	20	20	15.8
rs2083914	c.911G>T	1	20	5	19
rs11137040	c.2215-15C>G	1	20	5	26.7

*NA = not available

The list of Single Nucleotide Polimorphism of *CDK5RAP2* gene among children with microcephaly

rs Number	SNP	n	Present study Total	Present study %	European %
rs932974	c.-18T>C	4	20	20	100
rs932975	c.-2G>T	6	20	30	100
rs4836822	c.865G>C	6	20	30	3.3
rs2501727	c.2274T>C	6	20	30	1.71
rs4837768	c.4618G>C	2	20	10	2
rs35909061	c.4665G>T	1	20	5	0.07
rs77100552	c.5578T>C	1	20	5	NA
rs3750494	c.480A>C	3	20	15	3.4
rs34523498	c.3065G>A	1	20	5	4.2
rs3780679	c.3134G>C	1	20	5	99
rs6478475	c.4041G>A	1	20	5	0.9
rs4837768	c.4618G>C	4	20	20	20.8
rs3739822	c.5418C>T	1	20	5	1.7
rs37504945	c.480A>C	1	20	5	5.3

*NA = not available

The list of Single Nucleotide Polimorphism of *CENPJ* gene among children with microcephaly

rs Number	SNP	n	Present study Total	Present study %	European %
rs3742165	c.3042A>G	9	20	45	4.5
rs35498994	c.61A>G	11	20	55	1.2
rs9318917	c.3216+7A>G	4	20	20	5
rs9318911	c.4125A>G	3	20	15	100
rs35599563	c.2992-6delT	5	20	25	50
rs3742163	c.3367-12T>C	4	20	20	0

*NA = not available

The list of Single Nucleotide Polimorphism of *STIL* gene among children with microcephaly

SNP	cDNA	n	Present study Total	Present study %	European %
rs3125630	c.257C>T	16	20	80	5.4
rs10789505	c.1452C>G	5	20	25	8.8
rs13376679	c.2954A>G	3	20	15	3.4

*NA = not available

Appendix V. Ethical Clearance

**KOMITE ETIK PENELITIAN KESEHATAN
FAKULTAS KEDOKTERAN UNIVERSITAS DIPONEGORO
DAN RS Dr KARIADI SEMARANG**
Sekretariat: Kantor PD IV-Dekanat FK Undip
Jl. dr. Sutomo 18, Semarang
Telp/Fax.024-8446905

ETHICAL CLEARANCE
No. 91 / EC/FK/RSDK/2006

Komisi Etik Penelitian Kesehatan Fakultas Kedokteran Universitas Diponegoro/RS.Dr.Kariadi Semarang, setelah membaca dan menelaah proposal penelitian, dengan judul :

Deteksi Gen penyebab Retardasi Mental pada Populasi Retardasi Mental di Sekolah Luar Biasa (SLB) Se-Kotamadya Semarang

Peneliti Utama : dr. Farmaditya EP Mundhofir
Anggota Peneliti : 1. dr. Tri Indah Winarni,MSi.Med
2. dr. Asri Purwanti, M.Pd,Sp.A(K)

Penelitian : Dilaksanakan di 8 Sekolah Luar Biasa (SLB YPAC, SLB Swadaya, SLB Widya Bakti, SLB Pelita Ilmu, SLB Hj. Sumiyati, SLB Dharma Mulia, SLB Immanuel, SLB Negeri) Semarang dan Lab. Bioteknologi Kedokteran Undip Semarang.

Setuju untuk dilaksanakan, dengan memperhatikan prinsip-prinsip yang dinyatakan dalam Deklarasi Helsinki 1975, dan Pedoman Nasional Etik Penelitian Kesehatan (PNEPK) Departemen Kesehatan RI 2004.

Peneliti harus melampirkan 2 kopi lembar *Informed Consent* yang telah disetujui dan ditandatangani oleh peserta peneliti pada laporan penelitian.

Semarang, 27 Nopember 2006

Menyetujui,
Fakultas Kedokteran UNDIP
Dekan

Prof.dr.Kabalrachman,Sp-KK(K)
NIP.130 354 867

Komisi Etik Penelitian Kesehatan
Fakultas Kedokteran Undip/RS dr Kariadi
Ketua

Prof.Dr.dr.Tjahjono,Sp.PA(K)FIAC
NIP. 130 368 076