

REFERENCES

- Abuelo D. 2007. Microcephaly syndromes. *Sem Pediatr Neurol*. 14(3):118-27.
- Afadiyanti A. 2011. Mutation analysis of mental retardation and epilepsy in Indonesia. Master Thesis. Semarang: Diponegoro University.
- American Psychiatric Association. 1994. Diagnostic and Statistical Manual of Mental Disorders 4th Ed. Washington DC: American Psychiatric Association.
- Ashwal S, Michelson D, Plawner L. 2009. Practice Parameter: Evaluation of the child with microcephaly. *Neurology*. 73:887-96.
- Ayatollahi SMT, Shayan Z. Head circumference standards for school children of Shiraz, Iran. 2006. *J Trop Ped*. 52(6):406-10.
- Barker KT, Houlston RS. 2003. Overgrowth syndromes: is dysfunctional PI3-kinase signalling a unifying mechanism? *Eur J Hum Genet*. 11(9):665-70.
- Baxter PS, Rigby AS, Rotsaert MH, Wright I. 2009. Acquired microcephaly: causes, patterns, motor and IQ effects, and associated growth changes. *Pediatrics*. 124(2):590-5.
- Bell J, Bodmer D, Sistermans E, Ramsden SC. 2007. Practice guidelines for the Interpretation and Reporting of Unclassified Variants (UVs) in Clinical Molecular Genetics. *UK Clinical Molecular Genetics Society and the Dutch Society of Clinical Genetic Laboratory Specialists (Vereniging Klinisch Genetische Laboratorium specialisten; VKGL)*. Available from: <http://www.cmgs.org/BPGs/pdfs%20current%20bpgs/UV%20GUIDELINES%20ratified.pdf> [Downloaded 10 February 2010].

Cheong JL, Hunt RW, Anderson PJ, Howard K, Thompson DK, Wang HX, et al. 2008. Head growth in preterm infants: correlation with magnetic resonance imaging and neurodevelopmental outcome. *Pediatrics*. 121(6):e1534-40.

Delatycki MB, Danks A, Churchyard A, Zhou XP, Eng C. 2003. De novo germline *PTEN* mutation in a man with Lhermitte-Duclos disease which arose on the paternal chromosome and was transmitted to his child with polydactyly and Wormian bones. *J Med Genet*. 40(8):92.

Desir J, Cassart M, David P, Van Bogaert P, Abramowicz M. 2008. Primary microcephaly with *ASPM* mutation shows simplified cortical gyration with antero-posterior gradient pre- and post-natally. *Am J Med Genet. Part A*. 146A(11):1439-43.

Eigsti IM, Shapiro T. 2003. A systems neuroscience approach to autism: biological, cognitive, and clinical perspectives. *Ment Retard Dev Disabil Res Rev*. 9(3):205-15.

Faradz SMH. 1998. Fragile-X mental retardation and Fragile-X chromosomes in Indonesia Population. PhD Thesis. Sydney: University of New South Wales.

Faradz SMH. 2004. Laboratory Manual. Semarang: Molecular and Cytogenetics Unit Medical Biotechnology Laboratory Diponegoro University.

Field M, Tarpey PS, Smith R, Edkins S, O'Meara S, Stevens C et al. 2007. Mutations in the *BRWD3* gene cause X-linked mental retardation associated with macrocephaly. *Am J Hum Genet*. 81(2):367-74.

Frith C. 2003. What do imaging studies tell us about the neural basis of autism? *Novartis Found Symp*. 251:149-66

Goffin A, Hoefsloot LH, Bosgoed E, Swillen A, Fryns JP. 2001. *PTEN* mutation in a family with Cowden syndrome and autism. *Am J Med Genet.* 105(6):521-4.

Grantham R. 1974. Amino acid difference formula to help explain protein evolution. *Science.* 185(4154):862-4.

Groszer M, Erickson R, Scripture-Adams DD, Lesche R, Trumpp A, Zack JA, et al. 2001. Negative regulation of neural stem/progenitor cell proliferation by the Pten tumor suppressor gene in vivo. *Science.* 294(5549):2186-9.

Hassan MJ, Khurshid M, Azeem Z, John P, Ali G, Chishti MS, et al. 2007. Previously described sequence variant in *CDK5RAP2* gene in a Pakistani family with autosomal recessive primary microcephaly. *BMC Med Genet.* 8:58.

HGMD. 2011. The Human Gene Mutation Database. [online] Available at: <<http://www.hgmd.org/>> [Accessed 12 June 2011].

Hobert JA, Eng C. 2009. *PTEN* hamartoma tumor syndrome: An overview. *Genet Med.* 11(10):687– 694.

Jackson AP, Eastwood H, Bell SM, Adu J, Toomes C, Carr IM, et al. 2002. Identification of microcephalin, a protein implicated in determining the size of the human brain. *Am J Med Genet.* 71(1):136-42.

Jackson AP, McHale DP, Campbell DA, Jafri H, Rashid Y, Mannan J, et al. 1998. Primary autosomal recessive microcephaly (*MCPHI*) maps to chromosome 8p22-pter. *Am J Hum Genet.* 63(2):541-6.

Javierre BM, Fernandez AF, Richter J, Al-Shahrour F, Martin-Subero JI, Rodriguez - Ubueva J, et al. 2010. Changes in the pattern of DNA methylation associate with twin discordance in systemic lupus erythematosus. *Genome Res.* 20(2):170-9.

Kaindl AM, Passemard S, Kumar P, Kraemer N, Issa L, Zwirner A, et al. 2010. Many roads lead to primary autosomal recessive microcephaly. *Prog Neurobiol.* 90(3):363-83.

Karabiber H, Durmaz Y, Yakinci C, Kutlu O, Gumusalan Y, Yologlu S, Yalaz K. 2001. Head circumference measurement of urban children aged between 6 and 12 in Malatya, Turkey. *Brain and Dev.* 23: 801–804.

Komardjaja I. 2005. The place of people with intellectual disabilities in Bandung, Indonesia. *Health Place.* 11: 117-120.

Kumar A, Girimaji SC, Duvvari MR, Blanton SH. 2009. Mutations in *STIL*, encoding a pericentriolar and centrosomal protein, cause primary microcephaly. *Am J Med Genet.* 84(2):286-90.

Life Technologies. 2011. AmpFISTR Identifiler Direct PCR Amplification Kit User Guide. Carlsbad, California USA: Life Technologies Corporation. Available from: http://tools.invitrogen.com/content/sfs/manuals/cms_065522.pdf [Downloaded 4 November 2011].

Li J, Yen C, Liaw D, Podsypanina K, Bose S, Wang SI, et al. 1997. *PTEN*, a putative protein tyrosine phosphatase gene mutated in human brain, breast, and prostate cancer. *Science.* 275(5308):1943-7.

Machin G. 2009. Non-Identical Monozygotic Twins, Intermediate Twin Types, Zygosity Testing, and the Non-Random Nature of Monozygotic Twinning: A Review. *Am J Med Genet Part C Semin Med Genet.* 151C:110–127

Mahmood S, Ahmad W, Hassan MJ. 2011. Autosomal Recessive Primary Microcephaly (MCPH): clinical manifestations, genetic heterogeneity and mutation continuum. *Orphanet J Rare Dis.* 6:39.

Mathe E, Olivier M, Kato S, Ishioka C, Hainaut P, Tavtigian SV. 2006. Computational approaches for predicting the biological effect of p53 missense mutations: a comparison of three sequence analysis based methods. *Nucleic Acids Res.* 34(5):1317-25.

McCaffery P, Deutsch CK. 2005. Macrocephaly and the control of brain growth in autistic disorders. *Prog Neurobiol.* 77(1-2):38-56.

McCarthy EK, Goldstein B. Asymmetric spindle positioning. *Current opinion in cell biology.* 2006;18(1):79-85.

Miles JH, Hadden LL, Takahashi TN, Hillman RE. 2000. Head circumference is an independent clinical finding associated with autism. *Am J Med Genet.* 95(4):339-50.

Mundhofir FEP. 2008. Cytogenetics, Molecular and Clinical Studies among Mentally Retarded Individuals in Semarang. Master Thesis. Semarang: Diponegoro University.

National Healthcare Group Polyclinics. 2000. Anthropometric Study on Pre-School Children in Singapore. Singapore: Singapore Health Booklet.

National Survey SUSENAS Indonesia. 2000. Bangkok: Asia-Pacific Development Center on Disability (APCD) country report. Available from [http://www/gtid.net/countryreport/2003-CountryReport \(Indonesia2\).pdf](http://www/gtid.net/countryreport/2003-CountryReport%20(Indonesia2).pdf). [Accessed 10 November 2010].

Nellhaus G. 1968. Head circumference from birth to eighteen years. Practical composite international and interracial graphs. *Pediatrics.* 41(1):106-14.

Olney AH. 2007. Macrocephaly syndromes. *Semin Pediatr Neurol.* 14(3):128-35.

Orloff MS, Eng C. 2008. Genetic and phenotypic heterogeneity in the *PTEN* hamartoma tumour syndrome. *Oncogene.* 27: 5387–5397.

- Orrico A, Galli L, Buoni S, Orsi A, Vonella G, Sorrentino V. 2009. Novel *PTEN* mutations in neurodevelopmental disorders and macrocephaly. *Clin Genet.* 75(2):195-8.
- Page DT, Kuti OJ, Prestia C, Sur M. 2009. Haploinsufficiency for Pten and Serotonin transporter cooperatively influences brain size and social behavior. *Proc Natl Acad Sci USA.* 106(6):1989-94.
- Parisi MA, Dinulos MB, Leppig KA, Sybert VP, Eng C, Hudgins L. 2001. The spectrum and evolution of phenotypic findings in *PTEN* mutation positive cases of Bannayan-Riley-Ruvalcaba syndrome. *J Med Genet.* 38(1):52-8.
- Ramensky V, Bork P, Sunyaev S. 2002. Human non-synonymous SNPs: server and survey. *Nucleic Acids Res.* 30(17):3894-900.
- Roche AF, Mukherjee D, Guo SM, Moore WM. 1987. Head circumference reference data: birth to 18 years. *Pediatrics.* 79(5):706-12.
- Rollins JD, Collins JS, Holden KR. 2010. United States head circumference growth reference charts: birth to 21 years. *J Pediatr.* 156(6):907-13, 913.e1-2.
- Schouten JP, McElgunn CJ, Waaijer R, Zwiijnenburg D, Diepvens F, Pals G. 2002. Relative quantification of 40 nucleic acid sequences by multiplex ligation-dependent probe amplification. *Nucleic Acids Res.* 30(12):e57.
- Stoll C. 2001. Problems in the diagnosis of fragile X syndrome in young children are still present. *Am J Med Genet.* 100(2):110-5.
- Tavtigian SV, Deffenbaugh AM, Yin L, Judkins T, Scholl T, Samollow PB, et al. 2006. Comprehensive statistical study of 452 *BRCA1* missense substitutions with classification of eight recurrent substitutions as neutral. *J Med Genet.* 43(4):295-305.

The Genome Reference Consortium. 2011. Genome Reference Consortium. [online] Available at: <
<http://www.ncbi.nlm.nih.gov/projects/genome/assembly/grc/human/index.shtml>> [Accessed 12 June 2011].

Thornton GK, Woods CG. 2009. Primary microcephaly: do all roads lead to Rome? *Trends in genetics*. 25(11):501-10.

Tsuzaki S, Matsuo N, Saito M and Osano M. 1990. The head circumference growth curve for Japanese children between 0–4 years of age: comparison with Caucasian children and correlation with stature. *Annals of Human Biology*. 17(4): 297-303.

Varga EA, Pastore M, Prior T, Herman GE, McBride KL. 2009. The prevalence of *PTEN* mutations in a clinical pediatric study with autism spectrum disorders, developmental delay, and macrocephaly. *Genet Med*. 11(2):111-7.

Vissers LELM, de Ligt J, Gilissen C, Janssen I, Steehouwer M, de Vries P, et al. A *de novo* paradigm for mental retardation. *Nat genet*. 42(12): 1109-13.

Wang Z, Moulton J. SNPs, protein structure, and disease. 2001. *Human mutation*. 17(4):263-70.

Waternberg N, Silver S, Harel S, Lerman-Sagie T. 2002. Significance of Microcephaly Among Children With Developmental Disabilities. *J Child Neurol*. 17(2): 117-22.

Williams C, Dagli A, Battaglia A. 2008. Genetic disorders associated with macrocephaly. *Am J Med Genet. Part A*. 146A(15):2023-37.

Woods CG. 2004. Human microcephaly. *Curr Opin Neurobiol*. 14(1):112-7.

Woods CG, Bond J, Enard W. 2005. Autosomal recessive primary microcephaly (MCPH): a review of clinical, molecular, and evolutionary findings. *Am J Hum Genet.* 76(5):717-28.

Zhang X, Liu D, Lv S, Wang H, Zhong X, Liu B, et al. *CDK5RAP2* is required for spindle checkpoint function. 2009. *Cell Cycle.* 8(8): 1206-16.

Zori RT, Marsh DJ, Graham GE, Marliss EB, Eng C. 1998. Germline PTEN mutation in a family with Cowden syndrome and Bannayan-Riley-Ruvalcaba syndrome. *Am J Med Genet.* 80(4):399-402.