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Full Publication List

A. International Journal Publications (In chronological order)

1. Chan KW, Lee TL, **Chung BHY**, Yang X and Lau YL. Identification of five novel WASP mutations in Chinese Families with Wiskott-Aldrich Syndrome. *Human Mutation* 2002;20(2):151-152.
2. **Chung BHY**, Ha SY, Chan GCF, Chiang A, Lee TL, Ho HK, Lee CY, Luk CW and Lau YL. Klebsiella infection in patients with Thalassaemia. *Clinical Infectious Diseases* 2003;36: 575-579.
3. **Chung BHY**, Wong VCN and Ip. P. Prevalence of Neuromuscular Disease in Chinese Children: A Study in Southern China, *Journal of Child Neurology* 2003;18(3): 217-219.
4. Wong VCN and **Chung BHY**. Survey of Public Awareness, Attitudes, and Understanding toward Epilepsy in Hong Kong, *Epilepsia* 2003;44(2): 268-269.
5. Khong PL, Lam BCC, **Chung BHY**, Wong K.Y and Ooi GC. Diffusion-weighted MR imaging in neonatal nonketotic hyperglycinemia. *American Journal of Neuroradiology* 2003;24(6):1181-1183.
6. Lee WC, Leung JLS, Fung CW, **Chung BHY**, and Wong V. Spectrum of Anticonvulsant Hypersensitivity Syndrome: Controversy of Treatment. *Journal of Child Neurology* 2004;19(8):619-623.
7. **Chung BHY** and Lau YL. Reply to Lee: Severe bacterial infection in transfusion-dependent patients with thalassaemia major. *Clinical Infectious Diseases* 2004;38(8):1195.
8. Wong V, **Chung BHY** Hui S, Fong A, Lau C, Law B, Lo K, Shum T and Wong R. Cerebral Palsy – Correlation of Risk factors and Functional performance using Functional Independence Measure for Children (WeeFIM). *Journal of Child Neurology* 2004;19(11):887-893.
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10. Ip P, **Chung BHY**, Wong VCN and Chan KY. Subacute Sclerosing Panencephalitis in Children: Prevalence in South China. *Pediatric Neurology* 2004;31(1):46-51.
11. **Chung BHY**, Ip P, Wong VCN, Lo JYC and Harding B. Acute Fulminant SSPE with absent Measles and PCR studies in Cerebrospinal Fluid. *Pediatric Neurology* 2004;31(3): 222-224.
12. Khong PL, Zhou LJ, Ooi GC, **Chung BHY**, Cheung RTF and Wong VCN. The evaluation of Wallerian degeneration in chronic paediatric middle cerebral artery infarction using diffusion tensor MR imaging. *Cerebrovascular Diseases* 2004;18(3): 240-247.
13. Wong V, Hui S, Lee WC, Leung LSJ, Ho PKP, Lau WLC, Fung CW and **Chung BHY**. A Modified Screening Tool for Autism – Checklist for Autism in Toddlers (CHAT-23) for Chinese Children. *Pediatrics* 2004;114(2): e166-176.
14. **Chung BHY**, Wong V and Ip P. Spinal Muscular Atrophy: Survival Pattern and Functional Status. *Pediatrics* 2004;114(5):548-553.
15. Wong V, **Chung BHY** and Wong R. Pilot Survey of public awareness, attitudes and understanding towards epilepsy in Hong Kong. *Neurology Asia* 2004;9:21-27.

16. **Chung BHY**, Wat LCY, Wong V. Febrile Seizure in Southern Chinese children: Incidence and Recurrence. *Pediatric Neurology* 2006;34(2):121-126.
17. Wong VCN, **Chung BHY**, Li S, Goh W and Lee SL. Mutation of Gene in Spinal Muscular Atrophy Respiratory Distress Type I. *Pediatric Neurology* 2006;34(6):474-477.
18. Wong VCN, **Chung BHY**. Evaluating a Child with Partial Developmental Delay (ParDD), Global Developmental Delay (GDD)/Mental Retardation (MR): Clinical Expertise Based or Evidence-Based? *Current Pediatric Reviews* 2006;2(2):143-153.
19. **Chung BHY** and Wong VCN. Relationship between 5 Common Viruses and Febrile Seizure in Children. *Archives of Disease in Childhood* 2007;92:589-593.
20. **Chung BHY**, Tsang A.M.C. and Wong V.C.N. [Letter to the Editors] Neurologic complications in children hospitalized with influenza: comparison between USA and Hong Kong. *The Journal of Pediatrics* 2007;151(5):e17-e18.
21. **Chung BHY**, Ma ESK, Khong PL and Chan GCF. Inherited thrombophilic factors do not increase central venous catheter blockage in children with malignancy. *Pediatric Blood & Cancer* 2008;51:509-512.
22. **Chung BHY**, Lam ST, Tong TM, Lun KS, Chan DH, Fok SF, Or JS, Smith DK, Yang W and Lau YL. Identification of novel FBN1 and TGFBR2 mutations in 65 probands with Marfan syndrome or Marfan-like phenotypes. *American Journal of Medical Genetics* 2009;149 A(7):1452-9.
23. Fernandez BA, Roberts W, **Chung BHY**, Weksberg R, Meyn S, Szatmari P, Joseph-George AM, Mackay S, Whitten K, Noble B, Vardy C, Crosbie V, Luscombe S, Tucker E, Turner L, Marshall CR, Scherer SW. Phenotypic Spectrum Associated with De Novo and Inherited Deletions and Duplications at 16p11.2 in Individuals Ascertained for Diagnosis of Autism Spectrum Disorder. *Journal of Medical Genetics* 2010;47:195-203.
24. **Chung BHY**, Chu L, Forrest C, Silver R, Toi A, Blaser S, Viero S, Taylor G and Chitayat D. Fetal Forehead Hemangiopericytoma. Prenatal Diagnosis and Postnatal Outcome. *Ultrasound in Obstetrics and Gynecology* 2010;36(1):121-124.
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29. Wong VCN and **Chung BHY**. Value of clinical assessment in the diagnostic evaluation of Global Developmental Delay (GDD) using a Likelihood Ratio Model. *Brain & Development* 2011;33:548-557.
30. **Chung BHY**, Stavropoulos J, Marshall CR, Weksberg R, Scherer SW and Yoon G. 2q23 de novo microdeletion involving the MBD5 gene in a patient with developmental delay, postnatal microcephaly and distinct facial features. *American Journal of Medical Genetics (Part A)* 2011;155:424-429.

31. **Chung BHY**, Shaffer LG, Keating S, Johnson J, Casey B and Chitayat D. From VACTEL-H to Heterotaxy: Variable Expressivity of ZIC3 - Related Disorders. *American Journal of Medical Genetics (Part A)* 2011;155(5):1123-1128.
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36. Ho ACC, Liu APY, Lun KS, Tang WF, Chan KYK, Lau EYT, Tang MHY, Tan TY, **Chung BHY**. A newborn with a 790 kb chromosome 17p13.3 microduplication presenting with aortic stenosis, microcephaly and dysmorphic facial features – Is cardiac assessment necessary for all patients with 17p13.3 microduplication? *European Journal of Medical Genetics* 2012;55(12):758-762.
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41. Shek NW, Tan TY, Ding SC, **Chung BHY**, Lau ET, Tang MH. Prenatal Diagnosis of Agenesis of the Corpus Callosum and Cerebellar Vermian Hypoplasia Associated with a Microdeletion on Chromosome 1p32. *Case Report in Perinatal Medicine* 2013;2:39-45.
42. Tsang JS, Tong DKH, **Chung BHY**, Tang MHY, Lau ET, Chan GCF and Law SYK. Alport's syndrome: case of a giant esophageal tumor. *Esophagus* 2013;10:114-117.
43. **Chung BHY**, Tao VQ, Tso WWY. Copy number variation and autism: New insights and clinical implications. *Journal of the Formosan Medical Association* 2014;113:400-408.
44. Tao V, Chan KYK, Chu YWY, Mok GTK, Tan TY, Yang WL, Lee SL, Tang WF, Tso WWY, Lau ET, Kan ASY, Tang MH, Lau YL, **Chung BHY**. The clinical impact of chromosomal microarray on paediatric care in Hong Kong. *PLOS One* 2014;9(10):e109629.
45. Chan TK, Hui E, **Chung BHY**. A child born with Edward's syndrome: the legal and moral duty to accede to the request for parentage determination. *Journal of Medical Ethics* 2014;40:383-386.

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51. Luk HM, Wong VCH, Lo IFM, Chan KYK, Lau ET, Kan ASY, Tang MHY, Tang WF, She WMK, Chu Y, Sin WK, **Chung BHY**. A prenatal case of split-hand malformation associated with 17p13.3 triplication – A dilemma in genetic counseling, *European Journal of Medical Genetics* 2014;57:81-84.
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54. Zeng S., Yang J., **Chung BHY**, Lau YL, WL Yang. EFIN: predicting the functional impact of nonsynonymous single nucleotide polymorphisms in human genome. *BMC Genomics* 2014, 15:455-463.
55. Zhang J, Zhang Y, Yang J., Zhang L, Sun L, Pan HF, Hirankarn N, Ying D, Zeng S, Lee TL, Lau CS, Chan TM, Leung AMH, Mok CC, Wong SN, Lee KW, Ho MHK, Lee PPW, **Chung BHY**, Chong CY, Wong RWS, Mok MY, Wong WHS, Tong KL, Tse NKC, Li XP, Avihingsanon Y, Rianthavorn P, Deekajorndej T, Suphapeetiporn K, Shotelersuk V, Ying SKY, Fung SKS, Lai WM, Garcia-Barcelo MM, Cherny SS, Tam PKH, Cui Y, Sham PC, Yang S, Ye DQ, Zhang XJ, Lau YL. Three SNPs in chromosome 11q23.3 are independently associated with systemic lupus erythematosus in Asians. *Human Molecular Genetics* 2014;23(2):524-533.
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66. Kruszka P, Uwineza A, Mutesa L, Martinez AF, Abe Y, Zackai EH, Ganetzky R, **Chung BHY**, Stevenson RE, Adelstein RS, Ma X, Mullikin JC, Hong SK, Muenke M. Limb body wall complex, amniotic band sequence, or new syndrome caused by mutation in IQ Motif containing K (IQCK)? *Molecular Genetics & Genomic Medicine* 2015;3(5):424-432.
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