Contents

Cancer genetics
581 Constitutional de novo deletion CNV encompassing REST predisposes to diffuse hyperplastic perilobar nephroblastomatosis (HPLN)
Z Hyder, A Fairclough, M Groom, J Getty, E Alexander, E M van Veen, G Makin, C Sethuraman, V Tang, D G Evans, E R Maher, E R Woodward

Screening
587 Can population BRCA screening be applied in non-Ashkenazi Jewish populations? Experience in Macau population
Z Qiu, C N Kwok, H Dong, L Jiang, L Zhang, M Gao, H K Leong, L Wang, G Meng, S M Wang

Developmental defects
592 A novel homozygous variant in TRAPPC2L results in a neurodevelopmental disorder and disrupts TRAPP complex function
N Al-Jeri, V Oktar, P Ahinaz, M Milev, Z Valivullah, J Hegner, Y Sheng, W Chung, M Sacher, M Ganapathi

Diagnostics
602 Clinical phenotype and loss of the slow skeletal muscle troponin T in three new patients with recessive TNNT1 nemaline myopathy

Neurogenetics
609 Neurogenetic fetal akinesia and arthrogryposis: genetics, expanding genotype-phenotype and functional genomics

619 Frameshift mutation of Tnnn8a1 gene in mouse leads to an abnormal mitochondrial structure in the brain, correlating with hearing and memory impairment
P Song, Y Guan, X Chen, C Wu, A Qiao, H Jiang, Q Li, Y Huang, W Huang, M Xu, O Nientia, C Yuan, W Li, L Zhou, Z Xiao, S Pan, Y Hu

629 Systematic evaluation of ollfaction in patients with hereditary cystic kidney diseases/renal ciliopathies

637 Determinants of quality of life in Rett syndrome: new findings on associations with genotype
J Mendoza, J Downs, K Wong, H Leonard

Phenotypes
645 RAS41 phenotype overlaps with hereditary haemorrhagic telangiectasia: two case reports
M El Hajjouj, A Mekki, A Palmyre, M Eyries, F Soulmiér, I Bourgault Villada, A Oszanne, R Y Carlier, T Chintet

648 Biallelic variants in BRCA1 gene cause a recognisable phenotype within chromosomal instability syndromes reframed as BRCA1 deficiency