



## Contents

### Epigenetics

**1003** Histone modifications in Duchenne muscular dystrophy: pathogenesis insights and therapeutic implications

*Y Wei, Y Jiang, Y Lu, Q Hu*

### Cancer genetics

**1011** Genetic findings in people with schwannomas who do not meet clinical diagnostic criteria for *NF2*-related schwannomatosis

*M J Smith, C Perez-Becerril, M van der Meer, G J Burghel, S J Waller, M Carney, S Bunstone, K Fryer, N L Bowers, C L Hartley, P T Smith, S A Rutherford, S R Freeman, S K W Lloyd, O N Pathmanaban, A T King, D Halliday, C Duff, D G Evans*

**1016** Protein-truncating and rare missense variants in *ATM* and *CHEK2* and associations with cancer in UK Biobank whole-exome sequence data

*T K Mukhtar, N Wilcox, J Dennis, X Yang, M Naven, N Mavaddat, J R B Perry, E Gardner, D F Easton*

**1023** Novel germline *TP53* variant (p.(Phe109Ile)) confers high risk of cancer

*A Byrjalsen, U K Stoltze, C Lautrup, L L Christensen, T Mikkelsen, L Hjalgrim, J S Brok, C Dahl, K Schmiegelow, L Borgwardt, B R Diness, T V O Hansen, K A W Wadt*

**1026** A novel pathogenic germline chromosome 3 inversion in von Hippel-Lindau disease

*C D Vocke, C J Ricketts, S Pack, M Raffeld, S Hewitt, A P Lebensohn, L O'Brien, R Gautam, K Reynolds, L S Schmidt, K Choo, A Kenigsberg, S Gurram, E Y Chew, N Nilubol, P Chittaboina, M J Merino, M W Ball, W M Linehan*

### Neurogenetics

**1031** Christianson syndrome across the lifespan: genetic mutations and longitudinal study in children, adolescents, and adults

*B C Kavanaugh, J Elacio, C R Best, D G St Pierre, M F Pescosolido, Q Ouyang, J Biedermann, R S Bradley, J S Liu, R N Jones, E M Morrow*

## November 2024 Volume 61 Issue 11

**1040** Splice site variants in the canonical donor site of *MED13L* exon 7 lead to intron retention in patients with *MED13L* syndrome

*J Fauqueux, S BouSSION, C Thuillier, E Meurisse, D Lacombe, M Willems, A Piton, E Ait-Yahya, J Ghoumid, T Smol*

### Genotype-phenotype correlations

**1045** Biallelic variants in  $\alpha$ -tubulin isotypes cause female infertility characterised as recurrent preimplantation embryo arrest

*H Hu, X Wan, H Zhang, J Sun, F Meng, S Zhang, Y Gu, F Gong, H Zhao, G Lin, W Zheng*

**1053** Novel variants and genotype-phenotype correlation in a multicentre cohort of GNE myopathy in China

*K Jiao, J Zhang, Q Li, X Lv, Y Yu, B Zhu, H Zhong, X'en Yu, J Song, Q Ke, F Qian, X Luan, X Zhang, X Chang, L Wang, M Liu, J Dong, Z Zou, B Bu, H Jiang, L Liu, Y Li, D Yue, X Chang, Y Zheng, N Wang, M Gao, X Xia, N Cheng, T Wang, S-S Luo, J Xi, J Lin, J Lu, C Zhao, H Yang, P Lin, D Hong, Z Zhao, Z Wang, W Zhu*

**1062** Pathogenic *SATB2* missense variants affecting p.Gly392 have variable functional implications and result in diverse clinical phenotypes

*J den Hoed, H Hashimoto, M Khan, F Semmekrot, K A Bosanko, C Abe-Hatano, E Nakagawa, H Venselaar, N Quercia, L Chad, H Kurosaka, S Rondeau, S E Fisher, S Yamamoto, Y A Zarate*

### Phenotypes

**1068** Male-female phenotype correlation and dissociation related to mutations in the *ARX* gene

*C L Li*

This article has been made freely available

online under the BMJ Journals open

access scheme. See [http://authors.bmj.com/](http://authors.bmj.com/open-access/)

open-access/



This journal is a member of and subscribes to the principles of the Committee on Publication Ethics <http://publicationethics.org/>



When you have finished with this please recycle it

MCQs The online version of this article contains multiple choice questions hosted on BMJ Learning.