

Contents

Methods

- 289** CRISPR-Cas9: a new and promising player in gene therapy *L Xiao-Jie, X Hui-Ying, K Zun-Ping, C Jin-Lian, J Li-Juan*

MCQs

Telomere biology

- 297** The heritability of leucocyte telomere length dynamics *J B Hjelmberg, C Dalgård, S Möller, T Steenstrup, M Kimura, K Christensen, S O Kyvik, A Aviv*

Developmental defects

- 303** Disruption of *CLPB* is associated with congenital microcephaly, severe encephalopathy and 3-methylglutaconic aciduria *J-M Capo-Chichi, S Boissel, E Brustein, S Pickles, C Fallet-Bianco, C Nassif, L Patry, S Dobrzeniecka, M Liao, D Labuda, M E Samuels, F F Hamdan, C V Velde, G A Rouleau, P Drapeau, J L Michaud*

Genome-wide studies

- 312** Thyroid hormone resistance syndrome due to mutations in the thyroid hormone receptor α gene (*THRA*) *A Tytki-Szymańska, R Acuna-Hidalgo, M Krajewska-Walasek, A Lecka-Ambroziak, M Stehouwer, C Gilissen, H G Brunner, A Jurecka, A Różdżyńska-Świątkowska, A Hoischen, K H Chrzanowska*

New loci

- 317** A novel *APC* mutation defines a second locus for Cenani-Lenz syndrome *N Patel, E Faqeih, S Anazi, M Alfawareh, S M Wakil, D Colak, F S Alkuraya*
- 322** Positional mapping of *PRKD1*, *NRP1* and *PRDM1* as novel candidate disease genes in truncus arteriosus *R Shaheen, A A Hashem, M H Alghamdi, M Z Seidahmad, S M Wakil, K Dagrin, B Keavney, J Goodship, S Alyousif, F M Al-Habshan, K Alhussein, A Almoisheer, N Ibrahim, F S Alkuraya*

Genotype-phenotype correlations

- 330** De novo gain-of-function and loss-of-function mutations of *SCN8A* in patients with intellectual disabilities and epilepsy *M G Blanchard, M H Willemsen, J B Walker, S D Dib-Hajj, S G Waxman, M CJ Jongmans, T Kleefstra, B P van de Warrenburg, P Praamstra, J Nicolai, H G Ynema, R JM Bindels, M H Meisler, E-J Kamsteeg*
- 338** A founder *MYBPC3* mutation results in HCM with a high risk of sudden death after the fourth decade of life *C Calore, M De Bortoli, C Romualdi, A Lorenzon, A Angelini, C Basso, G Thiene, S Illiceto, A Rampazzo, P Melacini*

Cancer genetics

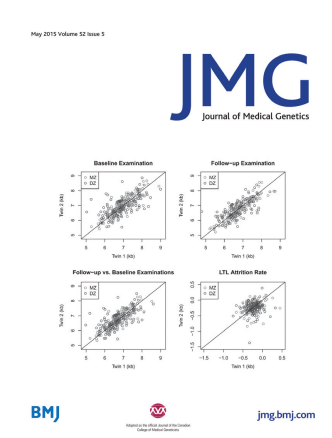
- 348** A homozygous *PMS2* founder mutation with an attenuated constitutional mismatch repair deficiency phenotype *L Li, N Hamel, K Baker, M J McGuffin, M Couillard, A Gologan, V A Marcus, B Chodirker, A Chudley, C Stefanovici, A Durandy, R A Hegele, B-J Feng, D E Goldgar, J Zhu, M De Rosa, S B Gruber, K Wimmer, B Young, G Chong, M D Tischkowitz, W D Foulkes*

Therapeutics

- 353** Ten-year outcome of enzyme replacement therapy with agalsidase beta in patients with Fabry disease *D P Germain, J Charrow, R J Desnick, N Guffon, J Kempf, R H Lachmann, R Lemay, G E Linthorst, S Packman, C R Scott, S Waldek, D G Warnock, N J Weinreb, W R Wilcox*

Obituary

- 360** A tribute to Margaret W. Thompson *L Siminovich, R Worton*



Cover credit: Twin correlations of telomere length metrics from *J B Hjelmberg et al*, pg 300.



Adopted as the official Journal of the Canadian College of Medical Geneticists

Receive regular table of contents by email. Register using this QR code.



This article has been chosen by the Editor to be of special interest or importance and is freely available online.



This article has been made freely available online under the BMJ Journals Open Access scheme. See <http://jmg.bmj.com/site/about/guidelines.xhtml#open>



COMMITTEE ON PUBLICATION ETHICS

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.