





Cover credit: Prediction of structural effects of a *GLI1* mutation and patient brain imaging, from Bölükbaşı *et al*, figure 4 (pg. 194) and supplementary figure S1.



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


 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://authors.bmj.com/open-access/>

 This journal is a member of and subscribes to the principles of the Committee on Publication Ethics <http://publicationethics.org/>
Member since 2008 JMG0012

 equator network

 recycle
When you have finished with this please recycle it

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

Contents

Therapeutics

- 143** Gene editing as a promising approach for respiratory diseases
Y Bai, Y Liu, Z Su, Y Ma, C Ren, R Zhao, H-L Ji

Genotype-phenotype correlations

- 150** Detection of heterozygous mutation in hook microtubule-tethering protein 1 in three patients with decapitated and decaudated spermatozoa syndrome
H Chen, Y Zhu, Z Zhu, E Zhi, K Lu, X Wang, F Liu, Z Li, W Xia
- 158** *P4HB* recurrent missense mutation causing Cole-Carpenter syndrome
M Balasubramanian, R Padidela, R C Pollitt, N J Bishop, M Z Mughal, A C Offiah, B E Wagner, J McCaughey, D J Stephens

Diagnostics

- 166** A false-carrier state for the c.579G>A mutation in the *NCF1* gene in Ashkenazi Jews
M De Boer, R Gavrieli, K van Leeuwen, H R Wolf, M Dushnitzki, Y Bar-Yosef, A Bar-Ziv, D Behar, S Lipitz, T E Miller, A T J Tool, T W Kuijpers, T K van den Berg, B Wolach, D Roos, E Pras

Cancer genetics

- 173** Contribution of de novo and mosaic *TP53* mutations to Li-Fraumeni syndrome
M Renaux-Petel, F Charbonnier, J-C Théry, P Fermeij, G Lienard, J Bou, S Coutant, M Vezain, E Kasper, S Fourneaux, S Manase, M Blanluet, B Leheup, L Mansuy, J Champigneulle, C Chappé, M Longy, N Sévenet, B Bressac-de Paillerets, L Guerrini-Rousseau, L Brugières, O Caron, J-C Sabourin, I Tournier, S Baert-Desurmont, T Frébourg, G Bougeard
- 181** Genome-wide association study identified copy number variants associated with sporadic colorectal cancer risk
L F Thean, Y S Low, M Lo, Y-Y Teo, W-P Koh, J-M Yuan, M H Chew, C L Tang, P Y Cheah

Phenotypes

- 189** Homozygous mutation in *CEP19*, a gene mutated in morbid obesity, in Bardet-Biedl syndrome with predominant postaxial polydactyly
E Yıldız Bölükbaşı, S Mumtaz, M Afzal, U Woehlbier, S Malik, A Tolun

Gametes

- 198** *DMC1* mutation that causes human non-obstructive azoospermia and premature ovarian insufficiency identified by whole-exome sequencing
W-B He, C-F Tu, Q Liu, L-L Meng, S-M Yuan, A-X Luo, F-S He, J Shen, W Li, J Du, C-G Zhong, G-X Lu, G Lin, L-Q Fan, Y-Q Tan

Epigenetics

- 205** Chromosomal rearrangements in the 11p15 imprinted region: 17 new 11p15.5 duplications with associated phenotypes and putative functional consequences
S Heide, S Chantot-Bastarud, B Keren, M D Harbison, S Azzi, S Rossignol, C Michot, M Lackmy-Port Lys, B Demeer, C Heinrichs, R S Newfield, P Sarda, L Van Maldergem, V Trifard, E Giabicani, J-P Siffroi, Y Le Bouc, I Netchine, F Brioude

Miscellaneous

- 214** Correction to: *Correction: FOXP1-related intellectual disability syndrome: a recognisable entity*