



Cover credit: Variant modelling of the the $\beta 5$ subunit of the guanine nucleotide-binding protein. See De Nittis *et al*, page 819.



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

Contents

Cancer genetics

789 Gene fusions in tumourigenesis with particular reference to ovarian cancer
MCQs
Y Zhou, M El-Bahrawy

796 Blood functional assay for rapid clinical interpretation of germline *TP53* variants
OPEN ACCESS

S Raad, M Rolain, S Coutant, C Derambure, R Lanos, F Charbonnier, J Bou, E Bouvignies, G Lienard, S Vasseur, M Farrell, O Ingster, S Baert Desurmont, E Kasper, G Bougeard, T Frébourg, I Tournier

807 Using chatbots to screen for heritable cancer syndromes in patients undergoing routine colonoscopy

B Heald, E Keel, J Marquard, C A Burke, M F Kalady, J M Church, D Liska, G Mankaney, K Hurley, C Eng

Functional genomics

815 Inhibition of G-protein signalling in cardiac dysfunction of intellectual developmental disorder with cardiac arrhythmia (IDDCA) syndrome
OPEN ACCESS

P De Nittis, S Eftymiou, A Sarre, N Guex, J Chrast, A Putoux, T Sultan, J Raza Alvi, Z ur Rahman, F Zafar, N Rana, F Rahman, N Anwar, S Maqbool, M S Zaki, J G Gleeson, D Murphy, H Galehdari, G Shariati, N Mazaheri, A Sedaghat, SYNAPS Study Group, G Lesca, N Chatriot, V Salpietro, M Christoforou, H Houlden, W F Simonds, T Pedrazzini, R Maroofian, A Reymond

December 2021 Volume 58 Issue 12

Genotype-phenotype correlations

832 Dilated cardiomyopathy caused by truncating titin variants: long-term outcomes, arrhythmias, response to treatment and sex differences
C R Vissing, T B Rasmussen, A M Dybro, M S Olesen, L N Pedersen, M Jensen, H Bundgaard, A H Christensen

Developmental defects

842 Prenatal features in Beckwith-Wiedemann syndrome and indications for prenatal testing
D Carli, C Bertola, S Cardaropoli, V P Ciuffreda, M Pieretto, G B Ferrero, A Mussa

Structural variation

850 CRISPR-Cas9/long-read sequencing approach to identify cryptic mutations in *BRCA1* and other tumour suppressor genes
OPEN ACCESS
T Walsh, S Casadei, K M Munson, M Eng, J B Mandell, S Gulsuner, M-C King

Clinical guidelines

853 Implementing gene curation for hereditary cancer susceptibility in Australia: achieving consensus on genes with clinical utility
E Tudini, A L Davidson, U Dressel, L Andrews, Y Antill, A Crook, M Field, M Gattas, R Harris, J Kirk, N Pachter, L Salmon, R Susman, S Townshend, A H Trainer, K M Tucker, G Mitchell, P A James, R L Ward, H Mar Fan, N K Poplawski, A B Spurdle



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/>



When you have finished with this please recycle it

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