### Contents

**Cancer genetics**

1035 Position statement of the International Society for Gastrointestinal Hereditary Tumours (InSIGHT) on APC 11307K and cancer risk  
L Valle, L H Katz, A Latchford, P Mur, V Moreno, I M Frayling, B Head, G Capellà, on behalf of the InSIGHT Council

1044 Ability of a polygenic risk score to refine colorectal cancer risk in Lynch syndrome  
N D ueñas, H Klifthammer, N Bonafaci, I Spier, A Mayer, E Hassanin, A Diez-Villanueva, V Moreno, M Pineda, C Maj, G Capellà, S Aretz, J Brunet

1052 Pancreatic cancer cluster region identified in BRCA2  
J S Chian, W Xu, S M Wang

1057 TPS3 c.455C>T p.(Pro152Leu) pathogenic variant is a lower risk allele with attenuated risks of breast cancer and sarcoma  
D G Evans, E F Harkness, E R Woodward

#### Genotype-phenotype correlations

1061 Analysis of Rotterdam Study cohorts confirms a previously identified RIPOR2 in-frame deletion as a prevalent genetic factor in phenotypically variable adult-onset hearing loss (DFNA21) in the Netherlands  
H M Velde, N C Homas, A Goedegebure, C P Lasting, R J E Pennings, H Kramer

1067 Pulmonary function and structure abnormalities in children and young adults with osteogenesis imperfecta point to intrinsic and extrinsic lung abnormalities  
B R G rocalco, M Hossain, S K Tabacchio, M X G Zuo, M Barton, A N Deng Do, J C Marinii

1076 Genotypes and phenotypes of DNMT1 encephalopathy  

### Neuroneurogenetics**

1105 HnRNP R strongly represses splicing of a critical exon associated with spinal muscular atrophy through binding to an exonic AU-rich element  
T Jang, R Qu, X Liu, Y Hou, L Wang, Y Hua

1116 Congenital mirror movements are associated with defective polymerisation of RAD51  
O Trouillat, P Dupaigne, M Dunoyer, M Doulatmi, M K Henn, S Frisman, A Riou, V Legros, G Chevrel, X Veaute, D Basso, C Fouquet, C Saint-Martin, A Ménetret, A Trembleau, I Dusart, C Dubacq, E Roze

### Novel disease loci

1127 Biallelic variants in DNA2 cause poikiloderma with congenital cataracts and severe growth failure reminiscent of Rothmund-Thomson syndrome  

1133 Exome sequencing links the SUMO protease SENP7 with fatal arthrogryposis multiplex congenita, early respiratory failure and neutropenia  