December 2022 Volume 59 Issue 12

1196 Prospective validation of the BOADICEA multifactorial breast cancer risk prediction model in a large prospective cohort study
X Yang, M Enkson, K Czene, A Lee, G Leslie, M Lush, J Wang, J Dennis, J Dorling, S Carvalho, N Mavaddat, J Simard, M K Schmidt, D F Easton, P Hall, A C Antoniou

1206 Enhancing the BOADICEA cancer risk prediction model to incorporate new data on RAD51C, RAD51D, BARDO1 updates to tumour pathology and cancer incidence

Contents

Genotype-phenotype correlations
1139 Association between SCNSA R2250 variant and dilated cardiomyopathy: potential role of intracellular pH and WNT/p-catenin pathway
J Hu, K Yang, Y Zhao, Z Wei, L Yang, G Cao, Y Wu, L Xu, S Xu, K Hu, A Sun, J Ge

Developmental defects
1151 Uncovering the burden of hidden ciliopathies in the 100 000 Genomes Project: a reverse phenotyping approach

Phenotypes
1171 Endocrine and behavioural features of Lowe syndrome and their potential molecular mechanisms
C Sena, G Iannello, A A Stawowski, K Daneluzzo, L Cheung, P B Agarwal, J N Hirschhorn, P Zeiler, C A De Luca, G Stratigopoulos, V V Thaker

Cancer genetics
1179 A digital pathway for genetic testing in UK NHS patients with cancer: BRCA-DIRECT randomised study internal pilot

Therapeutics
1165 The international Fragile X Premutation Registry: building a resource for research and clinical trial readiness

Neurogenetics
1219 TMPRSS3 expression is limited in spiral ganglion neurons: implication for successful cochlear implantation

Copy-number variation
1234 Exome sequencing as a first-tier test for copy number variant detection: retrospective evaluation and prospective screening in 2418 cases

Epigenetics
1241 CDKN1C hyperexpression in two patients with severe growth failure and microdeletions affecting the paternally inherited CDKN1C TSS-DMR
K Hana-Isono, K Yamazawa, S Taneaka, E Nishi, M Fukuji, M Kagami

Endocrine and behavioural features of Lowe syndrome and their potential molecular mechanisms. See page 1173.