

# CITATION REPORT

List of articles citing

**Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology**

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**Genetics in Medicine, 2015, 17, 405-24.**

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| 1437 | Evaluating the Calling Performance of a Rare Disease NGS Panel for Single Nucleotide and Copy Number Variants. <b>2017</b> , 21, 303-313                           | 7     |
| 1436 | Prediction of breast cancer risk based on flow-variant analysis of circulating peripheral blood B cells. <i>Genetics in Medicine</i> , <b>2017</b> , 19, 1071-1077 | 8.1 3 |

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| 1435 | Clinical laboratories collaborate to resolve differences in variant interpretations submitted to ClinVar. <i>Genetics in Medicine</i> , <b>2017</b> , 19, 1096-1104                                                        | 8.1 | 137 |
| 1434 | Novel mutations of PDGFRB cause primary familial brain calcification in Chinese families. <b>2017</b> , 62, 697-701                                                                                                        |     | 10  |
| 1433 | Nationwide genetic analysis for molecularly unresolved cystic fibrosis patients in a multiethnic society: implications for preconception carrier screening. <b>2017</b> , 5, 223-236                                       |     | 6   |
| 1432 | SDHA related tumorigenesis: a new case series and literature review for variant interpretation and pathogenicity. <b>2017</b> , 5, 237-250                                                                                 |     | 35  |
| 1431 | Principles and Recommendations for Standardizing the Use of the Next-Generation Sequencing Variant File in Clinical Settings. <b>2017</b> , 19, 417-426                                                                    |     | 13  |
| 1430 | Diagnostic Application of Targeted Next-Generation Sequencing of 80 Genes Associated with Disorders of Sexual Development. <b>2017</b> , 7, 44536                                                                          |     | 25  |
| 1429 | Genetics and Genomics of Congenital Heart Disease. <b>2017</b> , 120, 923-940                                                                                                                                              |     | 197 |
| 1428 | Common data elements for clinical research in mitochondrial disease: a National Institute for Neurological Disorders and Stroke project. <b>2017</b> , 40, 403-414                                                         |     | 11  |
| 1427 | De novo mtDNA point mutations are common and have a low recurrence risk. <b>2017</b> , 54, 73-83                                                                                                                           |     | 44  |
| 1426 | Two novel LHX3 mutations in patients with combined pituitary hormone deficiency including cervical rigidity and sensorineural hearing loss. <b>2017</b> , 17, 17                                                           |     | 9   |
| 1425 | A mild form of Stickler syndrome type II caused by mosaicism of COL11A1. <b>2017</b> , 60, 275-278                                                                                                                         |     | 9   |
| 1424 | Genomics, Endoscopy, and Control of Gastroesophageal Cancers: A Perspective. <b>2017</b> , 3, 359-366                                                                                                                      |     | 4   |
| 1423 | A CACNA1D mutation in a patient with persistent hyperinsulinaemic hypoglycaemia, heart defects, and severe hypotonia. <b>2017</b> , 18, 320-323                                                                            |     | 46  |
| 1422 | Guidelines for Validation of Next-Generation Sequencing-Based Oncology Panels: A Joint Consensus Recommendation of the Association for Molecular Pathology and College of American Pathologists. <b>2017</b> , 19, 341-365 |     | 310 |
| 1421 | Evaluating the quality of Marfan genotype-phenotype correlations in existing FBN1 databases. <i>Genetics in Medicine</i> , <b>2017</b> , 19, 772-777                                                                       | 8.1 | 21  |
| 1420 | IGHMBP2-related clinical and genetic features in a cohort of Chinese Charcot-Marie-Tooth disease type 2 patients. <b>2017</b> , 27, 193-199                                                                                |     | 7   |
| 1419 | The diagnostic yield of whole-exome sequencing targeting a gene panel for hearing impairment in The Netherlands. <b>2017</b> , 25, 308-314                                                                                 |     | 67  |
| 1418 | The Clinical Next-Generation Sequencing Database: A Tool for the Unified Management of Clinical Information and Genetic Variants to Accelerate Variant Pathogenicity Classification. <b>2017</b> , 38, 252-259             |     | 10  |

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| 1417 | Increased Identification of Candidates for High-Risk Breast Cancer Screening Through Expanded Genetic Testing. <b>2017</b> , 14, 561-568                                                                     | 19  |
| 1416 | Gitelman syndrome: consensus and guidance from a Kidney Disease: Improving Global Outcomes (KDIGO) Controversies Conference. <b>2017</b> , 91, 24-33                                                         | 148 |
| 1415 | Genetic compendium of 1511 human brains available through the UK Medical Research Council Brain Banks Network Resource. <b>2017</b> , 27, 165-173                                                            | 36  |
| 1414 | Vulvar and vaginal melanoma: A unique subclass of mucosal melanoma based on a comprehensive molecular analysis of 51 cases compared with 2253 cases of nongynecologic melanoma. <b>2017</b> , 123, 1333-1344 | 62  |
| 1413 | In silico analysis for predicting pathogenicity of five unclassified mitochondrial DNA mutations associated with mitochondrial cytopathies' phenotypes. <b>2017</b> , 60, 172-177                            | 7   |
| 1412 | Targeted full-scan LC-MS metabolomics: simultaneous quantitation of knowns and feature discovery provide the best of both worlds. <b>2017</b> , 9, 5-8                                                       | 3   |
| 1411 | Clinical characteristics and platelet phenotype in a family with RUNX1 mutated thrombocytopenia. <b>2017</b> , 58, 1963-1967                                                                                 | 7   |
| 1410 | A novel variant in RyR2 causes familial catecholaminergic polymorphic ventricular tachycardia. <b>2017</b> , 270, 173-177                                                                                    | 2   |
| 1409 | European Respiratory Society guidelines for the diagnosis of primary ciliary dyskinesia. <b>2017</b> , 49,                                                                                                   | 258 |
| 1408 | Issues and Challenges in Diagnostic Sequencing for Inherited Cardiac Conditions. <b>2017</b> , 63, 116-128                                                                                                   | 6   |
| 1407 | Atypical hemolytic uremic syndrome and C3 glomerulopathy: conclusions from a "Kidney Disease: Improving Global Outcomes" (KDIGO) Controversies Conference. <b>2017</b> , 91, 539-551                         | 335 |
| 1406 | Prevalence and Spectrum of Germline Cancer Susceptibility Gene Mutations Among Patients With Early-Onset Colorectal Cancer. <b>2017</b> , 3, 464-471                                                         | 335 |
| 1405 | Rapid Molecular Genetic Diagnosis with Next-Generation Sequencing in 46,XY Disorders of Sex Development Cases: Efficiency and Cost Assessment. <b>2017</b> , 87, 81-87                                       | 20  |
| 1404 | Phenotype and genotype analyses in seven families with dentinogenesis imperfecta or dentin dysplasia. <b>2017</b> , 23, 360-366                                                                              | 10  |
| 1403 | GJA1 gene variations in sudden unexplained nocturnal death syndrome in the Chinese Han population. <b>2017</b> , 270, 178-182                                                                                | 3   |
| 1402 | Design of a randomized controlled trial for genomic carrier screening in healthy patients seeking preconception genetic testing. <b>2017</b> , 53, 100-105                                                   | 22  |
| 1401 | The promises and challenges of exome sequencing in familial, non-syndromic congenital heart disease. <b>2017</b> , 230, 155-163                                                                              | 8   |
| 1400 | The novel homozygous KCNJ10 c.986T>C (p.(Leu329Pro)) variant is pathogenic for the SeSAME/EAST homologue in Malinois dogs. <b>2017</b> , 25, 222-226                                                         | 10  |



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| 1399 | A comprehensive global genotype-phenotype database for rare diseases. <b>2017</b> , 5, 66-75                                                                                                                                                                                   | 45  |
| 1398 | Variants of genes encoding collagens and matrix metalloproteinase system increased the risk of aortic dissection. <b>2017</b> , 60, 57-65                                                                                                                                      | 16  |
| 1397 | Standards and Guidelines for the Interpretation and Reporting of Sequence Variants in Cancer: A Joint Consensus Recommendation of the Association for Molecular Pathology, American Society of Clinical Oncology, and College of American Pathologists. <b>2017</b> , 19, 4-23 | 744 |
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| 1394 | mutations in Brazilian patients with sporadic pituitary adenomas: a single-center evaluation. <b>2017</b> , 6, 914-925                                                                                                                                                         | 8   |
| 1393 | Molecular Analysis Confirms that FKRP-Related Disorders are Underdiagnosed in Mexican Patients with Neuromuscular Diseases. <b>2017</b> , 48, 442-450                                                                                                                          | 2   |
| 1392 | Targeted Resequencing of Putative Growth-Related Genes Using Whole Exome Sequencing in Patients with Severe Primary IGF-I Deficiency. <b>2017</b> , 88, 408-417                                                                                                                | 7   |
| 1391 | Genetically Confirmed Familial Hypercholesterolemia in Patients With Acute Coronary Syndrome. <b>2017</b> , 70, 1732-1740                                                                                                                                                      | 79  |
| 1390 | Controversy and debate on clinical genomics sequencing-paper 2: clinical genome-wide sequencing: don't throw out the baby with the bathwater!. <b>2017</b> , 92, 7-10                                                                                                          |     |
| 1389 | Nationwide French Study of RET Variants Detected from 2003 to 2013 Suggests a Possible Influence of Polymorphisms as Modifiers. <b>2017</b> , 27, 1511-1522                                                                                                                    | 28  |
| 1388 | Novel Compound Heterozygous Mutations Cause Severe Fetal Microcephaly and Centriolar Lengthening. <b>2017</b> , 8, 282-293                                                                                                                                                     | 13  |
| 1387 | [Epilepsy-new diagnostic tools, old drugs? : Therapeutic consequences of epilepsy genetics]. <b>2017</b> , 88, 1385-1394                                                                                                                                                       | 3   |
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| 1384 | Vulnerability of mutation carriers to aripiprazole and trazodone exposure. <b>2017</b> , 58, 2139-2146                                                                                                                                                                         | 12  |
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| 1381 | High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. <b>2017</b> , 101, 664-685                                                                                                        | 214 |
| 1380 | Clinical testing with a panel of 25 genes associated with increased cancer risk results in a significant increase in clinically significant findings across a broad range of cancer histories. <b>2017</b> , 218-219, 58-68 | 38  |
| 1379 | Reassessing the significance of the PAH c.158G>A (p.Arg53His) variant in patients with hyperphenylalaninemia. <b>2017</b> , 30, 1211-1218                                                                                   | 7   |
| 1378 | Exome-wide Association Study Identifies GREB1L Mutations in Congenital Kidney Malformations. <b>2017</b> , 101, 789-802                                                                                                     | 36  |
| 1377 | P 43 A novel deletion in two exons of the SH3TC2 gene with mutation in the DPYD gene in Charcot-Marie-Tooth disease type 4C. <b>2017</b> , 128, e351-e352                                                                   |     |
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| 1374 | Favorable impact of allogeneic stem cell transplantation in patients with therapy-related myelodysplasia regardless of mutational status. <b>2017</b> , 102, 2030-2038                                                      | 17  |
| 1373 | Prenatal and early diagnosis of Chinese 3-M syndrome patients with novel pathogenic variants. <b>2017</b> , 474, 159-164                                                                                                    | 4   |
| 1372 | Genomic Triangulation and Coverage Analysis in Whole-Exome Sequencing-Based Molecular Autopsies. <b>2017</b> , 10,                                                                                                          | 12  |
| 1371 | Care in Specialized Centers and Data Sharing Increase Agreement in Hypertrophic Cardiomyopathy Genetic Test Interpretation. <b>2017</b> , 10,                                                                               | 27  |
| 1370 | Genomic Triangulation in Sudden Unexplained Death in the Young: The Way to Go?. <b>2017</b> , 10,                                                                                                                           | 1   |
| 1369 | Pathogenicity of Hypertrophic Cardiomyopathy Variants: A Path Forward Together. <b>2017</b> , 10,                                                                                                                           | 6   |
| 1368 | Knowledge for Precision Medicine: Mechanistic Reasoning and Methodological Pluralism. <b>2017</b> , 318, 1649-1650                                                                                                          | 35  |
| 1367 | Identification of twenty-nine novel germline unclassified variants of BRCA1 and BRCA2 genes in 1400 Italian individuals. <b>2017</b> , 36, 74-78                                                                            | 8   |
| 1366 | A novel mutation of VAPB in one Chinese familial amyotrophic lateral sclerosis pedigree and its clinical characteristics. <b>2017</b> , 264, 2387-2393                                                                      | 6   |
| 1365 | Repeat genetic testing with targeted capture sequencing in primary arrhythmia syndrome and cardiomyopathy. <b>2017</b> , 25, 1313-1323                                                                                      | 9   |
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| 1363 | Rapid Targeted Genomics in Critically Ill Newborns. <b>2017</b> , 140,                                                                                                                      | 69  |
| 1362 | Discrimination of Germline T790M Mutations in Plasma Cell-Free DNA Allows Study of Prevalence Across 31,414 Cancer Patients. <b>2017</b> , 23, 7351-7359                                    | 52  |
| 1361 | Functional analysis of novel DEAF1 variants identified through clinical exome sequencing expands DEAF1-associated neurodevelopmental disorder (DAND) phenotype. <b>2017</b> , 38, 1774-1785 | 15  |
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| 1359 | The challenges of implementing pharmacogenomic testing in the clinic. <b>2017</b> , 17, 567-577                                                                                             | 28  |
| 1358 | Heterozygous RFX6 protein truncating variants are associated with MODY with reduced penetrance. <b>2017</b> , 8, 888                                                                        | 57  |
| 1357 | Machine Learning and Rare Variant Adjudication in Type 1 Long QT Syndrome. <b>2017</b> , 10,                                                                                                | 2   |
| 1356 | Molecular and clinical profile of von Willebrand disease in Spain (PCM-EVW-ES): comprehensive genetic analysis by next-generation sequencing of 480 patients. <b>2017</b> , 102, 2005-2014  | 23  |
| 1355 | A Comparison of Whole Genome Sequencing to Multigene Panel Testing in Hypertrophic Cardiomyopathy Patients. <b>2017</b> , 10,                                                               | 45  |
| 1354 | Hypertrophic Cardiomyopathy Gene Testing: Go Big?. <b>2017</b> , 10,                                                                                                                        | 2   |
| 1353 | Clinical and molecular characterization of hereditary spastic paraplegias: A next-generation sequencing panel approach. <b>2017</b> , 383, 18-25                                            | 29  |
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| 1350 | Analysis and Annotation of Whole-Genome or Whole-Exome Sequencing Derived Variants for Clinical Diagnosis. <b>2017</b> , 95, 9.24.1-9.24.28                                                 | 9   |
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| 1348 | Clinical genetics and outcome of left ventricular non-compaction cardiomyopathy. <b>2017</b> , 38, 3449-3460                                                                                | 102 |
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| 1345 | Early repolarization syndrome caused by duplication of detected by next-generation sequencing. <b>2017</b> , 3, 574-578                                                                                           | 4   |
| 1344 | Massively parallel sequencing and targeted exomes in familial kidney disease can diagnose underlying genetic disorders. <b>2017</b> , 92, 1493-1506                                                               | 51  |
| 1343 | The prevalence of diseases caused by lysosome-related genes in a cohort of undiagnosed patients. <b>2017</b> , 13, 46-51                                                                                          | 12  |
| 1342 | Whole genome sequencing identifies etiology of recurrent male intrauterine fetal death. <b>2017</b> , 37, 1040-1045                                                                                               | 11  |
| 1341 | X-linked hypomyelination with spondylometaphyseal dysplasia (H-SMD) associated with mutations in AIFM1. <b>2017</b> , 18, 185-194                                                                                 | 28  |
| 1340 | Informed consent for next-generation nucleotide sequencing studies: Aiding communication between participants and investigators. <b>2017</b> , 1, 115-120                                                         | 5   |
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| 1338 | A mutational signature reveals alterations underlying deficient homologous recombination repair in breast cancer. <b>2017</b> , 49, 1476-1486                                                                     | 255 |
| 1337 | The Use of Variant Maps to Explore Domain-Specific Mutations of FGFR1. <b>2017</b> , 96, 1339-1345                                                                                                                | 4   |
| 1336 | The mutation-free embryo for in vitro fertilization selected by MALBAC-PGD resulted in a healthy live birth from a family carrying PKD 1 mutation. <b>2017</b> , 34, 1653-1658                                    | 11  |
| 1335 | Multigene Next-Generation Sequencing Panel Identifies Pathogenic Variants in Patients with Unknown Subtype of Epidermolysis Bullosa: Subclassification with Prognostic Implications. <b>2017</b> , 137, 2649-2652 | 23  |
| 1334 | High-Throughput Genetic Screening of 51 Pediatric Cataract Genes Identifies Causative Mutations in Inherited Pediatric Cataract in South Eastern Australia. <b>2017</b> , 7, 3257-3268                            | 13  |
| 1333 | Next-Generation Sequencing in Order to Better Characterize a BRCA Variant of Uncertain Significance. <b>2017</b> , 10, 634-637                                                                                    | 2   |
| 1332 | Molecular diversity of combined and complex dystonia: insights from diagnostic exome sequencing. <b>2017</b> , 18, 195-205                                                                                        | 25  |
| 1331 | The Case for Laboratory Developed Procedures: Quality and Positive Impact on Patient Care. <b>2017</b> , 4, 2374289517708309                                                                                      | 17  |
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| 1328 | A novel missense variant in the GLI3 zinc finger domain in a family with digital anomalies. <b>2017</b> , 173, 3221-3225                                                                                          |     |

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| 1327 | Variant Interpretation: Functional Assays to the Rescue. <b>2017</b> , 101, 315-325                                                                                                                              | 171 |
| 1326 | Functional precision cancer medicine-moving beyond pure genomics. <b>2017</b> , 23, 1028-1035                                                                                                                    | 161 |
| 1325 | The genetics of congenital heart disease—understanding and improving long-term outcomes in congenital heart disease: a review for the general cardiologist and primary care physician. <b>2017</b> , 29, 520-528 | 18  |
| 1324 | Unique genetic background and outcome of non-Caucasian Japanese probands with arrhythmogenic right ventricular dysplasia/cardiomyopathy. <b>2017</b> , 5, 639-651                                                | 10  |
| 1323 | Germline Mutations in Cancer Predisposition Genes are Frequent in Sporadic Sarcomas. <b>2017</b> , 7, 10660                                                                                                      | 37  |
| 1322 | Clinical and molecular analysis of 6 Chinese patients with isoleucine metabolism defects: identification of 3 novel mutations in the HSD17B10 and ACAT1 gene. <b>2017</b> , 32, 2063-2071                        | 4   |
| 1321 | Novel mutations of ATP7B gene in Wilson's disease patients of South Indian cohort. <b>2017</b> , 14, 114-118                                                                                                     |     |
| 1320 | Diagnostic Yield From 339 Epilepsy Patients Screened on a Clinical Gene Panel. <b>2017</b> , 77, 61-66                                                                                                           | 46  |
| 1319 | Frequent genes in rare diseases: panel-based next generation sequencing to disclose causal mutations in hereditary neuropathies. <b>2017</b> , 143, 507-522                                                      | 48  |
| 1318 | Arrhythmogenic Cardiomyopathy. <b>2017</b> , 121, 784-802                                                                                                                                                        | 167 |
| 1317 | Molecular genetic and clinical delineation of 22 patients with congenital hypogonadotropic hypogonadism. <b>2017</b> , 30, 1111-1118                                                                             | 11  |
| 1316 | A Wide and Specific Spectrum of Genetic Variants and Genotype-Phenotype Correlations Revealed by Next-Generation Sequencing in Patients with Left Ventricular Noncompaction. <b>2017</b> , 6,                    | 29  |
| 1315 | Targeted sequencing of 36 known or putative colorectal cancer susceptibility genes. <b>2017</b> , 5, 553-569                                                                                                     | 20  |
| 1314 | The defining DNA methylation signature of Kabuki syndrome enables functional assessment of genetic variants of unknown clinical significance. <b>2017</b> , 12, 923-933                                          | 43  |
| 1313 | Molecular and clinical spectra of FBXL4 deficiency. <b>2017</b> , 38, 1649-1659                                                                                                                                  | 29  |
| 1312 | Expanding the genetic heterogeneity of intellectual disability. <b>2017</b> , 136, 1419-1429                                                                                                                     | 74  |
| 1311 | Annotating pathogenic non-coding variants in genic regions. <b>2017</b> , 8, 236                                                                                                                                 | 77  |
| 1310 | A novel missense variant in the nuclear localization signal of POU4F3 causes autosomal dominant non-syndromic hearing loss. <b>2017</b> , 7, 7551                                                                | 12  |

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| 1309 | Prevalence of Rare Genetic Variations and Their Implications in NGS-data Interpretation. <b>2017</b> , 7, 9810                                                                                                     | 13  |
| 1308 | Interleukin-6, tumor necrosis factor-alpha and receptor activator of nuclear factor kappa ligand are elevated in hypertrophic gastric mucosa of pachydermoperiostosis. <b>2017</b> , 7, 9686                       | 3   |
| 1307 | Genetic screening confirms heterozygous mutations in ACAN as a major cause of idiopathic short stature. <b>2017</b> , 7, 12225                                                                                     | 37  |
| 1306 | Higher-than-expected population prevalence of potentially pathogenic germline TP53 variants in individuals unselected for cancer history. <b>2017</b> , 38, 1723-1730                                              | 24  |
| 1305 | Sequencing of transporter genes in cholestasis: We are still learning. <b>2017</b> , 67, 1132-1133                                                                                                                 | 3   |
| 1304 | Structural consequences of mutations associated with idiopathic restrictive cardiomyopathy. <b>2017</b> , 49, 1815-1829                                                                                            | 3   |
| 1303 | Optimizing genomic medicine in epilepsy through a gene-customized approach to missense variant interpretation. <b>2017</b> , 27, 1715-1729                                                                         | 91  |
| 1302 | Rapid whole-genome sequencing identifies a novel variant associated with West syndrome. <b>2017</b> , 3,                                                                                                           | 15  |
| 1301 | Mutation Detection in Patients With Advanced Cancer by Universal Sequencing of Cancer-Related Genes in Tumor and Normal DNA vs Guideline-Based Germline Testing. <b>2017</b> , 318, 825-835                        | 235 |
| 1300 | Two Novel Variants Affecting CDKL5 Transcript Associated with Epileptic Encephalopathy. <b>2017</b> , 21, 613-618                                                                                                  | 7   |
| 1299 | Congenital neutropenia in the era of genomics: classification, diagnosis, and natural history. <b>2017</b> , 179, 557-574                                                                                          | 77  |
| 1298 | Sudden infant death syndrome and inherited cardiac conditions. <b>2017</b> , 14, 715-726                                                                                                                           | 22  |
| 1297 | Identification of Novel Clinically Relevant Variants in 70 Southern Chinese patients with Thoracic Aortic Aneurysm and Dissection by Next-generation Sequencing. <b>2017</b> , 7, 10035                            | 16  |
| 1296 | Combined genetic analyses can achieve efficient diagnostic yields for subjects with Alagille syndrome and incomplete Alagille syndrome. <b>2017</b> , 106, 1817-1824                                               | 4   |
| 1295 | Combined study of ADAMTS13 and complement genes in the diagnosis of thrombotic microangiopathies using next-generation sequencing. <b>2017</b> , 1, 69-80                                                          | 11  |
| 1294 | Germline Mutations in Cancer Susceptibility Genes in a Large Series of Unselected Breast Cancer Patients. <b>2017</b> , 23, 6113-6119                                                                              | 103 |
| 1293 | Leveraging splice-affecting variant predictors and a minigene validation system to identify Mendelian disease-causing variants among exon-captured variants of uncertain significance. <b>2017</b> , 38, 1521-1533 | 22  |
| 1292 | Whole exome sequencing and DNA methylation analysis in a clinical amyotrophic lateral sclerosis cohort. <b>2017</b> , 5, 418-428                                                                                   | 8   |

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| 1291 | A deep intronic CLRN1 (USH3A) founder mutation generates an aberrant exon and underlies severe Usher syndrome on the Arabian Peninsula. <b>2017</b> , 7, 1411                     | 25 |
| 1290 | Effectiveness of levetiracetam in an acetazolamide-unresponsive patient with episodic ataxia type 2 by a novel CACNA1A nonsense mutation. <b>2017</b> , 24, e43-e44               | 7  |
| 1289 | High-throughput sequencing approaches for diagnosing hereditary bleeding and platelet disorders. <b>2017</b> , 15, 1262-1272                                                      | 29 |
| 1288 | Molecular diagnosis of autosomal dominant polycystic kidney disease. <b>2017</b> , 17, 885-895                                                                                    | 11 |
| 1287 | Findings of a 1303 Korean whole-exome sequencing study. <b>2017</b> , 49, e356                                                                                                    | 23 |
| 1286 | Clinical features and mutation spectrum in Chinese patients with CADASIL: A multicenter retrospective study. <b>2017</b> , 23, 707-716                                            | 25 |
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| 1284 | Early genetic testing for neonatal epilepsy: When, why, and how?. <b>2017</b> , 89, 880-881                                                                                       | 5  |
| 1283 | Rapidly Progressive White Matter Involvement in Early Childhood: The Expanding Phenotype of Infantile Onset Pompe?. <b>2018</b> , 39, 55-62                                       | 13 |
| 1282 | Biallelic COL3A1 mutations result in a clinical spectrum of specific structural brain anomalies and connective tissue abnormalities. <b>2017</b> , 173, 2534-2538                 | 17 |
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| 958 | Next generation sequencing analysis of patients with familial cervical artery dissection. <b>2017</b> , 2, 137-143                                                               | 13  |
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| 948 | Breast and Ovarian Cancer Penetrance Estimates Derived From Germline Multiple-Gene Sequencing Results in Women.. <b>2017</b> , 1, 1-12                                                | 45 |
| 947 | Efforts Toward Consensus Variant Interpretation by Commercial Laboratories. <b>2017</b> , 35, 1261-1262                                                                               | 3  |
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| 945 | When Should Genetic Testing Be Performed in Epilepsy Patients?. <b>2017</b> , 17, 16-22                                                                                               | 23 |
| 944 | The role of BRCA status on prognosis in patients with triple-negative breast cancer. <b>2017</b> , 8, 87151-87162                                                                     | 19 |
| 943 | Risk category system to identify pituitary adenoma patients with mutations. <b>2018</b> , 55, 254-260                                                                                 | 22 |
| 942 | Chromosomal aberrations and CNVs in twin fetuses with cardiovascular anomalies: Comparison between monozygotic diamniotic and dichorionic diamniotic twins. <b>2018</b> , 38, 318-327 | 7  |
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| 923 | Personalized cancer therapy-leveraging a knowledge base for clinical decision-making. <b>2018</b> , 4,                                                                                  | 37     |
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| 919 | A comprehensive evaluation of CHEK2 germline mutations in men with prostate cancer. <b>2018</b> , 78, 607-615                                                                           | 33     |
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| 915 | Statistical Validation of Rare Complement Variants Provides Insights into the Molecular Basis of Atypical Hemolytic Uremic Syndrome and C3 Glomerulopathy. <b>2018</b> , 200, 2464-2478 | 89     |
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| 913 | Human $\alpha$ -AMPK Mutations. <b>2018</b> , 1732, 581-619                                                                                                                                                                    | 2      |
| 912 | Disorders of spermatogenesis: Perspectives for novel genetic diagnostics after 20 years of unchanged routine. <b>2018</b> , 30, 12-20                                                                                          | 98     |
| 911 | Diagnostic strategies and genotype-phenotype correlation in a large Indian cohort of osteogenesis imperfecta. <b>2018</b> , 110, 368-377                                                                                       | 21     |
| 910 | Holt-Oram syndrome in two families diagnosed with left ventricular noncompaction and conduction disease. <b>2018</b> , 4, 146-151                                                                                              | 8      |
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| 908 | Molecular Diagnostics in Pathology: Time for a Next-Generation Pathologist?. <b>2018</b> , 142, 313-320                                                                                                                        | 25     |
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| 903 | Analysis of 17 genes detects mutations in 81% of 811 patients with lissencephaly. <i>Genetics in Medicine</i> , <b>2018</b> , 20, 1354-1364                                                                                    | 8.1 61 |
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| 897 | Similarities and differences between infantile and early childhood onset vanishing white matter disease. <b>2018</b> , 265, 1410-1418                                                                                          | 6      |
| 896 | Utility of DNA, RNA, Protein, and Functional Approaches to Solve Cryptic Immunodeficiencies. <b>2018</b> , 38, 307-319                                                                                                         | 21     |

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| 895 | Mutational spectrum of PTS gene and in silico pathological assessment of a novel variant in Mexico. <b>2018</b> , 40, 530-536                                                                              | 1   |
| 894 | Clinical and molecular insights into Glanzmann's thrombasthenia in China. <b>2018</b> , 94, 213-220                                                                                                        | 6   |
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| 892 | Analysis of state laws on informed consent for clinical genetic testing in the era of genomic sequencing. <b>2018</b> , 178, 81-88                                                                         | 7   |
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| 890 | Parental Mosaicism in "De Novo" Epileptic Encephalopathies. <b>2018</b> , 378, 1646-1648                                                                                                                   | 67  |
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| 883 | Diagnostic outcomes for genetic testing of 70 genes in 8565 patients with epilepsy and neurodevelopmental disorders. <b>2018</b> , 59, 1062-1071                                                           | 122 |
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| 881 | Genomic analysis identifies masqueraders of full-term cerebral palsy. <b>2018</b> , 5, 538-551                                                                                                             | 47  |
| 880 | Functional Characterization of Novel Phenylalanine Hydroxylase p.Gln226Lys Mutation Revealed Its Non-responsiveness to Tetrahydrobiopterin Treatment in Hepatoma Cellular Model. <b>2018</b> , 56, 533-541 | 1   |
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| 878 | MERTK mutation update in inherited retinal diseases. <b>2018</b> , 39, 887-913                                                                                                                             | 28  |

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| 877 | Genetic variant spectrum in 265 Chinese patients with hemophagocytic lymphohistiocytosis: Molecular analyses of PRF1, UNC13D, STX11, STXBP2, SH2D1A, and XIAP. <b>2018</b> , 94, 200-212 | 22     |
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| 875 | Mutations in plasmalemma vesicle-associated protein cause severe syndromic protein-losing enteropathy. <b>2018</b> , 55, 637-640                                                         | 14     |
| 874 | Homozygous missense MYBPC3 Pro873His mutation associated with increased risk for heart failure development in hypertrophic cardiomyopathy. <b>2018</b> , 5, 716-723                      | 7      |
| 873 | Characteristics of genomic alterations of lung adenocarcinoma in young never-smokers. <b>2018</b> , 143, 1696-1705                                                                       | 33     |
| 872 | Isolated Wolff-Parkinson-White syndrome in identical twins. <b>2018</b> , 4, 138-140                                                                                                     | 0      |
| 871 | Mutation Spectrum of GNE Myopathy in the Indian Sub-Continent. <b>2018</b> , 5, 85-92                                                                                                    | 12     |
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| 869 | Seven additional families with spondylocarpotarsal synostosis syndrome with novel biallelic deleterious variants in FLNB. <b>2018</b> , 94, 159-164                                      | 7      |
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| 866 | SCN1A variants associated with sudden infant death syndrome. <b>2018</b> , 59, e56-e62                                                                                                   | 19     |
| 865 | Biochemical and molecular characterisation of neurological Wilson disease. <b>2018</b> , 55, 587-593                                                                                     | 3      |
| 864 | Pathogenic Germline Variants in 10,389 Adult Cancers. <b>2018</b> , 173, 355-370.e14                                                                                                     | 342    |
| 863 | Autosomal dominant stapes fixation, syndactyly, and symphalangism in a family with NOG mutation: Long term follow-up on surgical treatment. <b>2018</b> , 108, 208-212                   | 1      |
| 862 | Hypertrophic cardiomyopathy. <b>2018</b> , 150, 434-442                                                                                                                                  | 1      |
| 861 | Expanding the phenome and variome of skeletal dysplasia. <i>Genetics in Medicine</i> , <b>2018</b> , 20, 1609-1616                                                                       | 8.1 20 |
| 860 | Prospective Study of Cancer Genetic Variants: Variation in Rate of Reclassification by Ancestry. <b>2018</b> , 110, 1059-1066                                                            | 26     |

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| 859 | Peripheral neuropathy in Tangier disease: A literature review and assessment. <b>2018</b> , 23, 88-98                                                                                 | 7  |
| 858 | Germline MLH1, MSH2 and MSH6 variants in Brazilian patients with colorectal cancer and clinical features suggestive of Lynch Syndrome. <b>2018</b> , 7, 2078-2088                     | 10 |
| 857 | Mutational analysis uncovers monogenic bone disorders in women with pregnancy-associated osteoporosis: three novel mutations in LRP5, COL1A1, and COL1A2. <b>2018</b> , 29, 1643-1651 | 25 |
| 856 | SD-OCT imaging as a valuable tool to support molecular genetic diagnostics of Usher syndrome type 1. <b>2018</b> , 22, 312-314.e3                                                     | 1  |
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| 854 | VarCards: an integrated genetic and clinical database for coding variants in the human genome. <b>2018</b> , 46, D1039-D1048                                                          | 86 |
| 853 | Prevalence and Progression of Late Gadolinium Enhancement in Children and Adolescents With Hypertrophic Cardiomyopathy. <b>2018</b> , 138, 782-792                                    | 40 |
| 852 | Targeted Next-generation Sequencing and Bioinformatics Pipeline to Evaluate Genetic Determinants of Constitutional Disease. <b>2018</b> ,                                             | 14 |
| 851 | A novel PIGA mutation in a Taiwanese family with early-onset epileptic encephalopathy. <b>2018</b> , 58, 52-54                                                                        | 8  |
| 850 | Long QT syndrome type 5-Lite: Defining the clinical phenotype associated with the potentially proarrhythmic p.Asp85Asn-KCNE1 common genetic variant. <b>2018</b> , 15, 1223-1230      | 13 |
| 849 | Identification of Misclassified ClinVar Variants via Disease Population Prevalence. <b>2018</b> , 102, 609-619                                                                        | 77 |
| 848 | Beyond BRCA: A Case Series Examining the Advent of Multigene Panel Testing. <b>2018</b> , 18, e431-e439                                                                               | 2  |
| 847 | Interplay Between Genetic Substrate, QTc Duration, and Arrhythmia Risk in Patients With Long QT Syndrome. <b>2018</b> , 71, 1663-1671                                                 | 76 |
| 846 | Targeted gene capture sequencing in diagnosis of dystonia patients. <b>2018</b> , 390, 36-41                                                                                          | 6  |
| 845 | Germline alterations in a consecutive series of acute myeloid leukemia. <b>2018</b> , 32, 2282-2285                                                                                   | 14 |
| 844 | Recognition of the polycistronic nature of human genes is critical to understanding the genotype-phenotype relationship. <b>2018</b> , 28, 609-624                                    | 34 |
| 843 | Inferring the effect of genomic variation in the new era of genomics. <b>2018</b> , 39, 756-773                                                                                       | 19 |
| 842 | 233rd ENMC International Workshop:: Clinical Trial Readiness for Calpainopathies, Naarden, The Netherlands, 15-17 September 2017. <b>2018</b> , 28, 540-549                           | 3  |

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| 840 | Exploring the feasibility and utility of exome-scale tumour sequencing in a clinical setting. <b>2018</b> , 48, 786-794                                                                                          |     | 3  |
| 839 | PINK1 p.K520RfsX3 mutation identified in a Chinese family with early-onset Parkinson's disease. <b>2018</b> , 676, 98-102                                                                                        |     | 3  |
| 838 | Variantes que mantienen el marco de lectura en el dominio Rod 1 proximal del gen FLNA se asocian con un predominio del fenotipo valvular. <b>2018</b> , 71, 545-552                                              |     | 3  |
| 837 | Antenatal screening and diagnosis of tuberous sclerosis complex by fetal echocardiography and targeted genomic sequencing. <b>2018</b> , 97, e0112                                                               |     | 9  |
| 836 | Muscle Weakness, Cardiomyopathy, and L-2-Hydroxyglutaric Aciduria Associated with a Novel Recessive SLC25A4 Mutation. <b>2019</b> , 43, 27-35                                                                    |     | 5  |
| 835 | The genetic characteristics of congenital hypothyroidism in China by comprehensive screening of 21 candidate genes. <b>2018</b> , 178, 623-633                                                                   |     | 46 |
| 834 | Periodic reanalysis of whole-genome sequencing data enhances the diagnostic advantage over standard clinical genetic testing. <b>2018</b> , 26, 740-744                                                          |     | 53 |
| 833 | A phenotype centric benchmark of variant prioritisation tools. <b>2018</b> , 3, 5                                                                                                                                |     | 24 |
| 832 | The NSIGHT1-randomized controlled trial: rapid whole-genome sequencing for accelerated etiologic diagnosis in critically ill infants. <b>2018</b> , 3, 6                                                         |     | 98 |
| 831 | Whole-exome sequencing for diagnosis of hereditary ichthyosis. <b>2018</b> , 32, 1022-1027                                                                                                                       |     | 12 |
| 830 | Genetic investigation of 93 families with microphthalmia or posterior microphthalmos. <b>2018</b> , 93, 1210-1222                                                                                                |     | 25 |
| 829 | Yield of the Genetic Test in Suspected Catecholaminergic Polymorphic Ventricular Tachycardia and Implications for Test Interpretation. <b>2018</b> , 11, e001424                                                 |     | 20 |
| 828 | The functional genomics laboratory: functional validation of genetic variants. <b>2018</b> , 41, 297-307                                                                                                         |     | 30 |
| 827 | Targeted gene panel and genotype-phenotype correlation in children with developmental and epileptic encephalopathy. <b>2018</b> , 141, 48-55                                                                     |     | 49 |
| 826 | Frequent mutations of RetNet genes in eoHM: Further confirmation in 325 probands and comparison with late-onset high myopia based on exome sequencing. <b>2018</b> , 171, 76-91                                  |     | 21 |
| 825 | A B3GALT6 variant in patient originally described as Al-Gazali syndrome and implicating the endoplasmic reticulum quality control in the mechanism of some BGalT6-pathway mutations. <b>2018</b> , 93, 1148-1158 |     | 7  |
| 824 | Genetics, Lifestyle, and Low-Density Lipoprotein Cholesterol in Young and Apparently Healthy Women. <b>2018</b> , 137, 820-831                                                                                   |     | 18 |



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| 823 | Assessment of the Clinical Relevance of BRCA2 Missense Variants by Functional and Computational Approaches. <b>2018</b> , 102, 233-248                             | 38  |
| 822 | Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. <b>2018</b> , 102, 309-320                                                              | 85  |
| 821 | Novel variant in Sp7/Osx associated with recessive osteogenesis imperfecta with bone fragility and hearing impairment. <b>2018</b> , 110, 66-75                    | 21  |
| 820 | Exome and genome sequencing in reproductive medicine. <b>2018</b> , 109, 213-220                                                                                   | 15  |
| 819 | Identification of co-occurrence in a patient with Dent's disease and ADA2-deficiency by exome sequencing. <b>2018</b> , 649, 23-26                                 | 5   |
| 818 | Paediatric genomics: diagnosing rare disease in children. <b>2018</b> , 19, 253-268                                                                                | 201 |
| 817 | Characteristics of Adrenal Masses in Familial Adenomatous Polyposis. <b>2018</b> , 61, 679-685                                                                     | 12  |
| 816 | Importance of complete phenotyping in prenatal whole exome sequencing. <b>2018</b> , 137, 175-181                                                                  | 39  |
| 815 | The first two confirmed sub-Saharan African families with germline TP53 mutations causing Li-Fraumeni syndrome. <b>2018</b> , 17, 607-613                          | 3   |
| 814 | A novel missense mutation in GIPC3 causes sensorineural hearing loss in an Iranian family revealed by targeted next-generation sequencing. <b>2018</b> , 108, 8-11 | 4   |
| 813 | Classification and Reporting of Potentially Proarrhythmic Common Genetic Variation in Long QT Syndrome Genetic Testing. <b>2018</b> , 137, 619-630                 | 43  |
| 812 | Multi-gene panel testing improves diagnosis and management of patients with hereditary anemias. <b>2018</b> , 93, 672-682                                          | 78  |
| 811 | Prediction of factor VIII inhibitor development in the SIPPET cohort by mutational analysis and factor VIII antigen measurement. <b>2018</b> , 16, 778-790         | 14  |
| 810 | Collagen VI-related myopathy: Expanding the clinical and genetic spectrum. <b>2018</b> , 58, 381-388                                                               | 10  |
| 809 | Prenatal diagnosis by chromosomal microarray analysis. <b>2018</b> , 109, 201-212                                                                                  | 128 |
| 808 | Consensus document on the implementation of next generation sequencing in the genetic diagnosis of hereditary cancer. <b>2018</b> , 151, 80.e1-80.e10              | 5   |
| 807 | Identification of rare genetic variants in Italian patients with dementia by targeted gene sequencing. <b>2018</b> , 66, 180.e23-180.e31                           | 15  |
| 806 | Screening for possible oligogenic pathogenesis in Chinese sporadic ALS patients. <b>2018</b> , 19, 419-425                                                         | 11  |

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| 805 | Identification of Five Novel Variants in Chinese Oculocutaneous Albinism by Targeted Next-Generation Sequencing. <b>2018</b> , 22, 252-258                                |     | 4  |
| 804 | Identification of a Novel Nonsense ASPM Mutation in a Large Consanguineous Pakistani Family Using Targeted Next-Generation Sequencing. <b>2018</b> , 22, 159-164          |     | 1  |
| 803 | SORL1 Variants in Familial Alzheimer's Disease. <b>2018</b> , 61, 1275-1281                                                                                               |     | 8  |
| 802 | ADA2 deficiency: Clonal lymphoproliferation in a subset of patients. <b>2018</b> , 141, 1534-1537.e8                                                                      |     | 43 |
| 801 | Neonatal Onset of Epilepsy of Infancy with Migrating Focal Seizures Associated with a Novel GABRB3 Variant in Monozygotic Twins. <b>2018</b> , 49, 204-208                |     | 10 |
| 800 | Advances in the Diagnosis and Management of Cystic Fibrosis in the Genomic Era. <b>2018</b> , 64, 898-908                                                                 |     | 8  |
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