Bruno Dallapiccola

List of Publications by Citations

560 papers	29,539 citations	78 h-index	152 g-index
572 ext. papers	33,250 ext. citations	5.9 avg, IF	6.22 L-index

#	Paper	IF	Citations
560	Hereditary early-onset Parkinson's disease caused by mutations in PINK1. *Science*, **2004**, 304, 1158-60	33.3	2586
559	LDL receptor-related protein 5 (LRP5) affects bone accrual and eye development. *Cell*, **2001**, 107, 513-23	36.2	1827
558	Gain-of-function RAF1 mutations cause Noonan and LEOPARD syndromes with hypertrophic cardiomyopathy. *Nature Genetics*, **2007**, 39, 1007-12	36.3	523
557	Gain-of-function SOS1 mutations cause a distinctive form of Noonan syndrome. *Nature Genetics*, **2007**, 39, 75-9	36.3	440
556	Mandibuloacral dysplasia is caused by a mutation in LMNA-encoding lamin A/C. *American Journal of Human Genetics*, **2002**, 71, 426-31	11	436
555	PINK1 mutations are associated with sporadic early-onset parkinsonism. *Annals of Neurology*, **2004**, 56, 336-41	9.4	397
554	Common variants at five new loci associated with early-onset inflammatory bowel disease. *Nature Genetics*, **2009**, 41, 1335-40	36.3	389
553	Mitochondrial import and enzymatic activity of PINK1 mutants associated to recessive parkinsonism. *Human Molecular Genetics*, **2005**, 14, 3477-92	5.6	376
552	Gut microbiota profiling of pediatric nonalcoholic fatty liver disease and obese patients unveiled by an integrated meta-omics-based approach. *Hepatology*, **2017**, 65, 451-464	11.2	354
551	Mutations in the pericentrin (PCNT) gene cause primordial dwarfism. *Science*, **2008**, 319, 816-9	33.3	325
550	Cystinuria caused by mutations in rBAT, a gene involved in the transport of cystine. *Nature Genetics*, **1994**, 6, 420-5	36.3	322
549	Grouping of multiple-lentigines/LEOPARD and Noonan syndromes on the PTPN11 gene. *American Journal of Human Genetics*, **2002**, 71, 389-94	11	321
548	Mutations in CEP290, which encodes a centrosomal protein, cause pleiotropic forms of Joubert syndrome. *Nature Genetics*, **2006**, 38, 623-5	36.3	320
547	Parkes Weber syndrome, vein of Galen aneurysmal malformation, and other fast-flow vascular anomalies are caused by RASA1 mutations. *Human Mutation*, **2008**, 29, 959-65	4.7	315
546	Transmembrane 6 superfamily member 2 gene variant disentangles nonalcoholic steatohepatitis from cardiovascular disease. *Hepatology*, **2015**, 61, 506-14	11.2	311
545	Mutations in INPP5E, encoding inositol polyphosphate-5-phosphatase E, link phosphatidyl inositol signaling to the ciliopathies. *Nature Genetics*, **2009**, 41, 1032-6	36.3	310
544	Development and validation of a multidimensional prognostic index for one-year mortality from comprehensive geriatric assessment in hospitalized older patients. *Rejuvenation Research*, **2008**, 11, 151-61	2.6	293

543	Mutation of SHOC2 promotes aberrant protein N-myristoylation and causes Noonan-like syndrome with loose anagen hair. *Nature Genetics*, **2009**, 41, 1022-6	36.3	291
542	The origin of the major cystic fibrosis mutation (delta F508) in European populations. *Nature Genetics*, **1994**, 7, 169-75	36.3	284
541	Mutations in a new gene in Ellis-van Creveld syndrome and Weyers acrodental dysostosis. *Nature Genetics*, **2000**, 24, 283-6	36.3	267
540	Deletion of KDM6A, a histone demethylase interacting with MLL2, in three patients with Kabuki syndrome. *American Journal of Human Genetics*, **2012**, 90, 119-24	11	258
539	Joubert Syndrome and related disorders. *Orphanet Journal of Rare Diseases*, **2010**, 5, 20	4.2	239
538	A restricted spectrum of NRAS mutations causes Noonan syndrome. *Nature Genetics*, **2010**, 42, 27-9	36.3	232
537	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. *American Journal of Human Genetics*, **2017**, 101, 664-685	11	214
536	Germline BRAF mutations in Noonan, LEOPARD, and cardiofaciocutaneous syndromes: molecular diversity and associated phenotypic spectrum. *Human Mutation*, **2009**, 30, 695-702	4.7	213
535	Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. *Nature Genetics*, **2010**, 42, 619-25	36.3	210
534	Additive effects of genetic variation in dopamine regulating genes on working memory cortical activity in human brain. *Journal of Neuroscience*, **2006**, 26, 3918-22	6.6	198
533	Further delineation of deletion 1p36 syndrome in 60 patients: a recognizable phenotype and common cause of developmental delay and mental retardation. *Pediatrics*, **2008**, 121, 404-10	7.4	197
532	Prenatal Diagnosis of Myotonic Dystrophy Using Fetal DNA Obtained from Maternal Plasma. *Clinical Chemistry*, **2000**, 46, 301-302	5.5	191
531	Mutations in lectin complement pathway genes COLEC11 and MASP1 cause 3MC syndrome. *Nature Genetics*, **2011**, 43, 197-203	36.3	190
530	Congenital heart diseases in children with Noonan syndrome: An expanded cardiac spectrum with high prevalence of atrioventricular canal. *Journal of Pediatrics*, **1999**, 135, 703-6	3.6	189
529	Germline missense mutations affecting KRAS Isoform B are associated with a severe Noonan syndrome phenotype. *American Journal of Human Genetics*, **2006**, 79, 129-35	11	183
528	Heterozygous germline mutations in the CBL tumor-suppressor gene cause a Noonan syndrome-like phenotype. *American Journal of Human Genetics*, **2010**, 87, 250-7	11	179
527	Mutations in PYCR1 cause cutis laxa with progeroid features. *Nature Genetics*, **2009**, 41, 1016-21	36.3	178
526	Leopard syndrome. *Orphanet Journal of Rare Diseases*, **2008**, 3, 13	4.2	174

525	Survival motor neuron gene transcript analysis in muscles from spinal muscular atrophy patients. *Biochemical and Biophysical Research Communications*, **1995**, 213, 342-8	3.4	169
524	Mutations in ANKRD11 cause KBG syndrome, characterized by intellectual disability, skeletal malformations, and macrodontia. *American Journal of Human Genetics*, **2011**, 89, 289-94	11	161
523	AHI1 is required for photoreceptor outer segment development and is a modifier for retinal degeneration in nephronophthisis. *Nature Genetics*, **2010**, 42, 175-80	36.3	152
522	Natural gene-expression variation in Down syndrome modulates the outcome of gene-dosage imbalance. *American Journal of Human Genetics*, **2007**, 81, 252-63	11	152
521	The human gut microbiota: a dynamic interplay with the host from birth to senescence settled during childhood. *Pediatric Research*, **2014**, 76, 2-10	3.2	144
520	Genetic susceptibility to nonsteroidal anti-inflammatory drug-related gastroduodenal bleeding: role of cytochrome P450 2C9 polymorphisms. *Gastroenterology*, **2007**, 133, 465-71	13.3	134
519	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. *Nature Genetics*, **2015**, 47, 661-7	36.3	128
518	Mutant Pink1 induces mitochondrial dysfunction in a neuronal cell model of Parkinson's disease by disturbing calcium flux. *Journal of Neurochemistry*, **2009**, 108, 1561-74	6	128
517	Searching for psoriasis susceptibility genes in Italy: genome scan and evidence for a new locus on chromosome 1. *Journal of Investigative Dermatology*, **1999**, 112, 32-5	4.3	125
516	Gerodermia osteodysplastica is caused by mutations in SCYL1BP1, a Rab-6 interacting golgin. *Nature Genetics*, **2008**, 40, 1410-2	36.3	121
515	Prevalence and clinical significance of cardiovascular abnormalities in patients with the LEOPARD syndrome. *American Journal of Cardiology*, **2007**, 100, 736-41	3	121
514	The INSL3-LGR8/GREAT ligand-receptor pair in human cryptorchidism. *Journal of Clinical Endocrinology and Metabolism*, **2003**, 88, 4273-9	5.6	120
513	CEP290 mutations are frequently identified in the oculo-renal form of Joubert syndrome-related disorders. *American Journal of Human Genetics*, **2007**, 81, 104-13	11	118
512	Multidimensional Prognostic Index based on a comprehensive geriatric assessment predicts short-term mortality in older patients with heart failure. *Circulation: Heart Failure*, **2010**, 3, 14-20	7.6	115
511	NF1 gene mutations represent the major molecular event underlying neurofibromatosis-Noonan syndrome. *American Journal of Human Genetics*, **2005**, 77, 1092-101	11	115
510	Mutations of ZFPM2/FOG2 gene in sporadic cases of tetralogy of Fallot. *Human Mutation*, **2003**, 22, 372-7	4.7	113
509	AHI1 gene mutations cause specific forms of Joubert syndrome-related disorders. *Annals of Neurology*, **2006**, 59, 527-34	9.4	111
508	Assignment of a locus for autosomal dominant idiopathic scoliosis (IS) to human chromosome 17p11. *Human Genetics*, **2002**, 111, 401-4	6.3	111

507	The search for south European cystic fibrosis mutations: identification of two new mutations, four variants, and intronic sequences. *Genomics*, **1991**, 10, 193-200	4.3	111
506	Anatomic patterns of conotruncal defects associated with deletion 22q11. *Genetics in Medicine*, **2001**, 3, 45-8	8.1	110
505	Guidelines for the appropriate use of genetic tests in infertile couples. *European Journal of Human Genetics*, **2002**, 10, 303-12	5.3	109
504	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. *American Journal of Human Genetics*, **2018**, 102, 175-187	11	108
503	Mediterranean diet and health: food effects on gut microbiota and disease control. *International Journal of Molecular Sciences*, **2014**, 15, 11678-99	6.3	107
502	Genotypes and phenotypes of Joubert syndrome and related disorders. *European Journal of Medical Genetics*, **2008**, 51, 1-23	2.6	106
501	Diversity, parental germline origin, and phenotypic spectrum of de novo HRAS missense changes in Costello syndrome. *Human Mutation*, **2007**, 28, 265-72	4.7	104
500	LEOPARD syndrome: clinical diagnosis in the first year of life. *American Journal of Medical Genetics, Part A*, **2006**, 140, 740-6	2.5	102
499	Familial transposition of the great arteries caused by multiple mutations in laterality genes. *Heart*, **2010**, 96, 673-7	5.1	100
498	22q11 deletions in isolated and syndromic patients with tetralogy of Fallot. *Human Genetics*, **1995**, 95, 479-82	6.3	99
497	Reference ranges of HOMA-IR in normal-weight and obese young Caucasians. *Acta Diabetologica*, **2016**, 53, 251-60	3.9	98
496	Germline and somatic NF1 mutations in sporadic and NF1-associated malignant peripheral nerve sheath tumours. *Journal of Pathology*, **2009**, 217, 693-701	9.4	98
495	Correlation between cardiac involvement and CTG trinucleotide repeat length in myotonic dystrophy. *Journal of the American College of Cardiology*, **1995**, 25, 239-45	15.1	98
494	Hennekam syndrome can be caused by FAT4 mutations and be allelic to Van Maldergem syndrome. *Human Genetics*, **2014**, 133, 1161-7	6.3	95
493	Activating mutations in RRAS underlie a phenotype within the RASopathy spectrum and contribute to leukaemogenesis. *Human Molecular Genetics*, **2014**, 23, 4315-27	5.6	95
492	A functional variant of the adipocyte glycerol channel aquaporin 7 gene is associated with obesity and related metabolic abnormalities. *Diabetes*, **2007**, 56, 1468-74	0.9	95
491	Cayler cardiofacial syndrome and del 22q11: part of the CATCH22 phenotype. *American Journal of Medical Genetics Part A*, **1994**, 53, 303-4		93
490	Recurrent microdeletion at 17q12 as a cause of Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome: two case reports. *Orphanet Journal of Rare Diseases*, **2009**, 4, 25	4.2	90

489	p.Arg1809Cys substitution in neurofibromin is associated with a distinctive NF1 phenotype without neurofibromas. *European Journal of Human Genetics*, **2015**, 23, 1068-71	5.3	89
488	Distinguishing the four genetic causes of Jouberts syndrome-related disorders. *Annals of Neurology* , **2005**, 57, 513-9	9.4	89
487	The functional Q84R polymorphism of mammalian Tribbles homolog TRB3 is associated with insulin resistance and related cardiovascular risk in Caucasians from Italy. *Diabetes*, **2005**, 54, 2807-11	0.9	89
486	Complete transposition of the great arteries: patterns of congenital heart disease in familial precurrence. *Circulation*, **2001**, 104, 2809-14	16.7	88
485	Congenital heart defects in Kabuki syndrome. *American Journal of Medical Genetics Part A*, **2001**, 100, 269-74		87
484	SOS1 mutations in Noonan syndrome: molecular spectrum, structural insights on pathogenic effects, and genotype-phenotype correlations. *Human Mutation*, **2011**, 32, 760-72	4.7	82
483	Park6-linked parkinsonism occurs in several european families. *Annals of Neurology*, **2002**, 51, 14-18	9.4	81
482	Gut Microbiota Markers in Obese Adolescent and Adult Patients: Age-Dependent Differential Patterns. *Frontiers in Microbiology*, **2018**, 9, 1210	5.7	78
481	Genetically determined interaction between the dopamine transporter and the D2 receptor on prefronto-striatal activity and volume in humans. *Journal of Neuroscience*, **2009**, 29, 1224-34	6.6	78
480	Mutations in PVRL4, encoding cell adhesion molecule nectin-4, cause ectodermal dysplasia-syndactyly syndrome. *American Journal of Human Genetics*, **2010**, 87, 265-73	11	78
479	Description, nomenclature, and mapping of a novel cerebello-renal syndrome with the molar tooth malformation. *American Journal of Human Genetics*, **2003**, 73, 663-70	11	78
478	A homozygous GJA1 gene mutation causes a Hallermann-Streiff/ODDD spectrum phenotype. *Human Mutation*, **2004**, 23, 286	4.7	77
477	Copy-number variations involving the IHH locus are associated with syndactyly and craniosynostosis. *American Journal of Human Genetics*, **2011**, 88, 70-5	11	76
476	Deletion of a 5-cM region at chromosome 8p23 is associated with a spectrum of congenital heart defects. *Circulation*, **2000**, 102, 432-7	16.7	76
475	UFD1L, a developmentally expressed ubiquitination gene, is deleted in CATCH 22 syndrome. *Human Molecular Genetics*, **1997**, 6, 259-65	5.6	75
474	Atypical deletions suggest five 22q11.2 critical regions related to the DiGeorge/velo-cardio-facial syndrome. *European Journal of Human Genetics*, **1999**, 7, 903-9	5.3	74
473	Epistasis between dopamine regulating genes identifies a nonlinear response of the human hippocampus during memory tasks. *Biological Psychiatry*, **2008**, 64, 226-34	7.9	73
472	An ATG repeat in the 3'-untranslated region of the human resistin gene is associated with a decreased risk of insulin resistance. *Journal of Clinical Endocrinology and Metabolism*, **2002**, 87, 4403-6	5.6	73

471	Keppen-Lubinsky syndrome is caused by mutations in the inwardly rectifying K+ channel encoded by KCNJ6. *American Journal of Human Genetics*, **2015**, 96, 295-300	11	72
470	MKS3/TMEM67 mutations are a major cause of COACH Syndrome, a Joubert Syndrome related disorder with liver involvement. *Human Mutation*, **2009**, 30, E432-42	4.7	72
469	Incidence of chromosome abnormalities and clinical significance of karyotype in de novo acute myeloid leukemia. *Cancer Genetics and Cytogenetics*, **1993**, 67, 28-34		72
468	Expression of DeltaF508 CFTR in normal mouse lung after site-specific modification of CFTR sequences by SFHR. *Gene Therapy*, **2001**, 8, 961-5	4	71
467	Type 2 deiodinase polymorphism (threonine 92 alanine) predicts L-thyroxine dose to achieve target thyrotropin levels in thyroidectomized patients. *Journal of Clinical Endocrinology and Metabolism*, **2008**, 93, 910-3	5.6	70
466	Comparison of occurrence of genetic syndromes in ventricular septal defect with pulmonic stenosis (classic tetralogy of Fallot) versus ventricular septal defect with pulmonic atresia. *American Journal of Cardiology*, **1996**, 77, 1375-6	3	70
465	Gut Microbiota Dysbiosis as Risk and Premorbid Factors of IBD and IBS Along the Childhood-Adulthood Transition. *Inflammatory Bowel Diseases*, **2016**, 22, 487-504	4.5	69
464	Changes in CpG islands promoter methylation patterns during ductal breast carcinoma progression. *Cancer Epidemiology Biomarkers and Prevention*, **2009**, 18, 2694-700	4	69
463	Mutations of the Nogo-66 receptor (RTN4R) gene in schizophrenia. *Human Mutation*, **2004**, 24, 534-5	4.7	69
462	Cardiac malformations in patients with oral-facial-skeletal syndromes: clinical similarities with heterotaxia. *American Journal of Medical Genetics Part A*, **1999**, 84, 350-6		69
461	BRF1 mutations alter RNA polymerase III-dependent transcription and cause neurodevelopmental anomalies. *Genome Research*, **2015**, 25, 155-66	9.7	68
460	Mutation spectrum of MLL2 in a cohort of Kabuki syndrome patients. *Orphanet Journal of Rare Diseases*, **2011**, 6, 38	4.2	68
459	Erythrocytes-mediated delivery of dexamethasone in steroid-dependent IBD patients-a pilot uncontrolled study. *American Journal of Gastroenterology*, **2005**, 100, 1370-5	0.7	66
458	Low doses of dexamethasone constantly delivered by autologous erythrocytes slow the progression of lung disease in cystic fibrosis patients. *Blood Cells, Molecules, and Diseases*, **2004**, 33, 57-63	2.1	66
457	Haploinsufficiency of the NOTCH1 Receptor as a Cause of Adams-Oliver Syndrome With Variable Cardiac Anomalies. *Circulation: Cardiovascular Genetics*, **2015**, 8, 572-581		65
456	Fine mapping of the PSORS4 psoriasis susceptibility region on chromosome 1q21. *Journal of Investigative Dermatology*, **2001**, 116, 728-30	4.3	65
455	A restricted spectrum of mutations in the SMAD4 tumor-suppressor gene underlies Myhre syndrome. *American Journal of Human Genetics*, **2012**, 90, 161-9	11	64
454	The Multidimensional Prognostic Index (MPI), based on a comprehensive geriatric assessment predicts short- and long-term mortality in hospitalized older patients with dementia. *Journal of Alzheimer's Disease*, **2009**, 18, 191-9	4.3	64

453	Phylogenetic and Metabolic Tracking of Gut Microbiota during Perinatal Development. *PLoS ONE*, **2015**, 10, e0137347	3.7	63
452	Prostaglandin transporter mutations cause pachydermoperiostosis with myelofibrosis. *Human Mutation*, **2012**, 33, 1175-81	4.7	63
451	Benchmarks for cystic fibrosis carrier screening: a European consensus document. *Journal of Cystic Fibrosis*, **2010**, 9, 165-78	4.1	63
450	The multidimensional prognostic index predicts short- and long-term mortality in hospitalized geriatric patients with pneumonia. *Journals of Gerontology - Series A Biological Sciences and Medical Sciences*, **2009**, 64, 880-7	6.4	63
449	Deficiency for the ubiquitin ligase UBE3B in a blepharophimosis-ptosis-intellectual-disability syndrome. *American Journal of Human Genetics*, **2012**, 91, 998-1010	11	62
448	Sex differences in the association of apolipoprotein E and angiotensin-converting enzyme gene polymorphisms with healthy aging and longevity: a population-based study from Southern Italy. *Journals of Gerontology - Series A Biological Sciences and Medical Sciences*, **2006**, 61, 918-23	6.4	62
447	Incidence and expression of the N1303K mutation of the cystic fibrosis (CFTR) gene. *Human Genetics*, **1992**, 89, 653-8	6.3	62
446	Common fragile sites: their prevalence in subjects with constitutional and acquired chromosomal instability. *American Journal of Medical Genetics Part A*, **1987**, 27, 471-82		61
445	Joubert syndrome and related disorders. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2013**, 113, 1879-88	3	60
444	Familial recurrence of congenital heart disease: an overview and review of the literature. *European Journal of Pediatrics*, **2007**, 166, 111-6	4.1	60
443	Associated cardiac anomalies in isolated and syndromic patients with tetralogy of Fallot. *American Journal of Cardiology*, **1996**, 77, 505-8	3	60
442	Erythrocyte-mediated delivery of dexamethasone in patients with mild-to-moderate ulcerative colitis, refractory to mesalamine: a randomized, controlled study. *American Journal of Gastroenterology*, **2008**, 103, 2509-16	0.7	59
441	A 4-polymorphism risk score predicts steatohepatitis in children with nonalcoholic fatty liver disease. *Journal of Pediatric Gastroenterology and Nutrition*, **2014**, 58, 632-6	2.8	58
440	Genetics of pediatric obesity. *Pediatrics*, **2012**, 130, 123-33	7.4	58
439	SHOX duplications found in some cases with type I Mayer-Rokitansky-Kuster-Hauser syndrome. *Genetics in Medicine*, **2010**, 12, 634-40	8.1	58
438	Interstitial 22q13 deletions: genes other than SHANK3 have major effects on cognitive and language development. *European Journal of Human Genetics*, **2008**, 16, 1301-10	5.3	58
437	PINK1 heterozygous rare variants: prevalence, significance and phenotypic spectrum. *Human Mutation*, **2008**, 29, 565	4.7	58
436	Atrioventricular canal defect without Down syndrome: A heterogeneous malformation. *American Journal of Medical Genetics Part A*, **1999**, 85, 140-146		58

435	Evidence for interaction between psoriasis-susceptibility loci on chromosomes 6p21 and 1q21. *American Journal of Human Genetics*, **1999**, 65, 1798-800	11	58
434	Sequence-specific modification of genomic DNA by small DNA fragments. *Journal of Clinical Investigation*, **2003**, 112, 637-41	15.9	58
433	Kabuki syndrome: clinical and molecular diagnosis in the first year of life. *Archives of Disease in Childhood*, **2015**, 100, 158-64	2.2	57
432	Spectrum of MEK1 and MEK2 gene mutations in cardio-facio-cutaneous syndrome and genotype-phenotype correlations. *European Journal of Human Genetics*, **2009**, 17, 733-40	5.3	57
431	Novel TMEM67 mutations and genotype-phenotype correlates in meckelin-related ciliopathies. *Human Mutation*, **2010**, 31, E1319-31	4.7	57
430	Nonrandom association of atrioventricular canal and del (8p) syndrome. *American Journal of Medical Genetics Part A*, **1992**, 42, 424-7		56
429	Targeted correction of a defective selectable marker gene in human epithelial cells by small DNA fragments. *Molecular Therapy*, **2001**, 3, 178-85	11.7	55
428	Parental origin of chromosome 4p deletion in Wolf-Hirschhorn syndrome. *American Journal of Medical Genetics Part A*, **1993**, 47, 921-4		55
427	POPDC1(S201F) causes muscular dystrophy and arrhythmia by affecting protein trafficking. *Journal of Clinical Investigation*, **2016**, 126, 239-53	15.9	55
426	Biallelic Mutations in TBCD, Encoding the Tubulin Folding Cofactor D, Perturb Microtubule Dynamics and Cause Early-Onset Encephalopathy. *American Journal of Human Genetics*, **2016**, 99, 962-973	11	55
425	Copy-Number Variation of the Glucose Transporter Gene SLC2A3 and Congenital Heart Defects in the 22q11.2 Deletion Syndrome. *American Journal of Human Genetics*, **2015**, 96, 753-64	11	54
424	Analysis of Clinically Relevant Single-Nucleotide Polymorphisms by Use of Microelectronic Array Technology. *Clinical Chemistry*, **2002**, 48, 2124-2130	5.5	52
423	Gut Microbiota Profiling and Gut-Brain Crosstalk in Children Affected by Pediatric Acute-Onset Neuropsychiatric Syndrome and Pediatric Autoimmune Neuropsychiatric Disorders Associated With Streptococcal Infections. *Frontiers in Microbiology*, **2018**, 9, 675	5.7	51
422	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. *Human Mutation*, **2015**, 36, 1080-7	4.7	51
421	Whole gene deletion and splicing mutations expand the PINK1 genotypic spectrum. *Human Mutation*, **2007**, 28, 98	4.7	51
420	Analysis of the epsilon-sarcoglycan gene in familial and sporadic myoclonus-dystonia: evidence for genetic heterogeneity. *Movement Disorders*, **2003**, 18, 1047-51	7	51
419	Epilepsy with auditory features: a LGI1 gene mutation suggests a loss-of-function mechanism. *Annals of Neurology*, **2003**, 53, 396-9	9.4	51
418	Transposition of the great arteries associated with deletion of chromosome 22q11. *American Journal of Cardiology*, **1995**, 75, 95-8	3	51

417	Assessing the role of DRD5 and DYT1 in two different case-control series with primary blepharospasm. *Movement Disorders*, **2007**, 22, 162-6	7	50
416	cDNA characterization and chromosomal mapping of two human homologues of the Drosophila dishevelled polarity gene. *Human Molecular Genetics*, **1996**, 5, 953-8	5.6	50
415	The TRIB3 Q84R polymorphism and risk of early-onset type 2 diabetes. *Journal of Clinical Endocrinology and Metabolism*, **2009**, 94, 190-6	5.6	49
414	Spinal muscular atrophy genotyping by gene dosage using multiple ligation-dependent probe amplification. *Neurogenetics*, **2006**, 7, 269-76	3	49
413	Deletion 22q11 in patients with interrupted aortic arch. *American Journal of Cardiology*, **1999**, 84, 360-1, A9	3	49
412	Recurrent Mutations in the Basic Domain of TWIST2 Cause Ablepharon Macrostomia and Barber-Say Syndromes. *American Journal of Human Genetics*, **2015**, 97, 99-110	11	48
411	Diagnosis of Noonan syndrome and related disorders using target next generation sequencing. *BMC Medical Genetics*, **2014**, 15, 14	2.1	48
410	Novel and recurrent EVC and EVC2 mutations in Ellis-van Creveld syndrome and Weyers acrofacial dyostosis. *European Journal of Medical Genetics*, **2013**, 56, 80-7	2.6	48
409	Cardiac defects, morbidity and mortality in patients affected by RASopathies. CARNET study results. *International Journal of Cardiology*, **2017**, 245, 92-98	3.2	48
408	Early-life gut microbiota under physiological and pathological conditions: the central role of combined meta-omics-based approaches. *Journal of Proteomics*, **2012**, 75, 4580-7	3.9	48
407	Genotype and cardiovascular phenotype correlations with TBX1 in 1,022 velo-cardio-facial/DiGeorge/22q11.2 deletion syndrome patients. *Human Mutation*, **2011**, 32, 1278-89	4.7	48
406	An excess of chromosome 1 breakpoints in male infertility. *European Journal of Human Genetics*, **2004**, 12, 993-1000	5.3	48
405	Deletion 8p syndrome **1998**, 75, 534-536		47
404	RAS signaling in colorectal carcinomas through alteration of RAS, RAF, NF1, and/or RASSF1A. *Neoplasia*, **2008**, 10, 680-6, 2 p following 686	6.4	47
403	Reliability of DHPLC in mutational screening of beta-globin (HBB) alleles. *Human Mutation*, **2002**, 19, 287-95	4.7	47
402	Majewski osteodysplastic primordial dwarfism type II (MOPD II) complicated by stroke: clinical report and review of cerebral vascular anomalies. *American Journal of Medical Genetics, Part A*, **2005** , 139, 212-5	2.5	47
401	Gut microbiota signatures in cystic fibrosis: Loss of host CFTR function drives the microbiota enterophenotype. *PLoS ONE*, **2018**, 13, e0208171	3.7	47
400	The Pitt-Hopkins syndrome: report of 16 new patients and clinical diagnostic criteria. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1536-45	2.5	46

399	COMT Val158Met polymorphism predicts negative symptoms response to treatment with olanzapine in schizophrenia. *Schizophrenia Research*, **2007**, 95, 253-5	3.6	46
398	Genomic instability and increased expression of BUB1B and MAD2L1 genes in ductal breast carcinoma. *Cancer Letters*, **2007**, 254, 298-307	9.9	45
397	DiGeorge subtypes of nonsyndromic conotruncal defects: evidence against a major role of TBX1 gene. *European Journal of Human Genetics*, **2003**, 11, 349-51	5.3	45
396	Mutations in KCNK4 that Affect Gating Cause a Recognizable Neurodevelopmental Syndrome. *American Journal of Human Genetics*, **2018**, 103, 621-630	11	45
395	High-resolution SNP arrays in mental retardation diagnostics: how much do we gain?. *European Journal of Human Genetics*, **2010**, 18, 178-85	5.3	44
394	Three patients with oculo-auriculo-vertebral spectrum and microdeletion 22q11.2. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 2860-4	2.5	44
393	A novel PTPN11 gene mutation bridges Noonan syndrome, multiple lentigines/LEOPARD syndrome and Noonan-like/multiple giant cell lesion syndrome. *European Journal of Human Genetics*, **2004**, 12, 1069-72	5.3	44
392	Interaction between PPARgamma2 variants and gender on the modulation of body weight. *Obesity*, **2008**, 16, 1467-70	8	43
391	Non-apolipoprotein E and apolipoprotein E genetics of sporadic Alzheimer's disease. *Ageing Research Reviews*, **2009**, 8, 214-36	12	42
390	Array-based comparative genomic hybridization in early-stage mycosis fungoides: recurrent deletion of tumor suppressor genes BCL7A, SMAC/DIABLO, and RHOF. *Genes Chromosomes and Cancer*, **2008**, 47, 1067-75	5	42
389	Mapping a dominant form of multinodular goiter to chromosome Xp22. *American Journal of Human Genetics*, **2000**, 67, 1004-7	11	42
388	PINK1 homozygous W437X mutation in a patient with apparent dominant transmission of parkinsonism. *Movement Disorders*, **2006**, 21, 1265-7	7	41
387	Genotype-phenotype analysis and natural history of left ventricular hypertrophy in LEOPARD syndrome. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 620-8	2.5	40
386	Human UDP-galactose 4'-epimerase (GALE) gene and identification of five missense mutations in patients with epimerase-deficiency galactosemia. *Molecular Genetics and Metabolism*, **1998**, 63, 26-30	3.7	40
385	Centric fission of chromosome no. 4 in the mother of two patients with trisomy 4p. *Human Genetics*, **1976**, 31, 121-5	6.3	40
384	The internet user profile of Italian families of patients with rare diseases: a web survey. *Orphanet Journal of Rare Diseases*, **2013**, 8, 76	4.2	39
383	Congenital heart defects in patients with oculo-auriculo-vertebral spectrum (Goldenhar syndrome). *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1815-9	2.5	39
382	Molecular detection of novel WFS1 mutations in patients with Wolfram syndrome by a DHPLC-based assay. *Human Mutation*, **2003**, 21, 622-9	4.7	39

381	Novel and recurrent mutations in the NF1 gene in Italian patients with neurofibromatosis type 1. *Human Mutation*, **2004**, 23, 629	4.7	38
380	Novel locus for autosomal dominant pure hereditary spastic paraplegia (SPG19) maps to chromosome 9q33-q34. *Annals of Neurology*, **2002**, 51, 681-5	9.4	38
379	NF1 gene analysis based on DHPLC. *Human Mutation*, **2003**, 21, 171-2	4.7	38
378	Familial blepharospasm is inherited as an autosomal dominant trait and relates to a novel unassigned gene. *Movement Disorders*, **2003**, 18, 207-12	7	38
377	Specific variants in WDR35 cause a distinctive form of Ellis-van Creveld syndrome by disrupting the recruitment of the EvC complex and SMO into the cilium. *Human Molecular Genetics*, **2015**, 24, 4126-37	5.6	37
376	Identification of TBX5 mutations in a series of 94 patients with Tetralogy of Fallot. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 3100-7	2.5	37
375	Congenital heart defects in molecularly proven Kabuki syndrome patients. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2912-2922	2.5	37
374	Application of MLPA assay to characterize unsolved □globin gene rearrangements. *Blood Cells, Molecules, and Diseases*, **2011**, 46, 139-44	2.1	37
373	Screening of mutations in the CFTR gene in 1195 couples entering assisted reproduction technique programs. *European Journal of Human Genetics*, **2005**, 13, 959-64	5.3	37
372	(CTG)n triplet mutation and phenotype manifestations in myotonic dystrophy patients. *Biochemical Medicine and Metabolic Biology*, **1993**, 50, 85-92		37
371	Foodomics as part of the host-microbiota-exposome interplay. *Journal of Proteomics*, **2016**, 147, 3-20	3.9	37
370	TBCE Mutations Cause Early-Onset Progressive Encephalopathy with Distal Spinal Muscular Atrophy. *American Journal of Human Genetics*, **2016**, 99, 974-983	11	37
369	Mutation screening of the DYT6/THAP1 gene in Italy. *Movement Disorders*, **2009**, 24, 2424-7	7	36
368	A combined analytical approach reveals novel EXT1/2 gene mutations in a large cohort of Italian multiple osteochondromas patients. *Genes Chromosomes and Cancer*, **2007**, 46, 470-7	5	36
367	Association of dopamine D4 receptor (DRD4) exon III repeat polymorphism with temperament in 3-year-old infants. *Neurogenetics*, **2003**, 4, 207-12	3	36
366	Atrioventricular canal and 8p- syndrome. *American Journal of Medical Genetics Part A*, **1993**, 47, 437-8		35
365	Aicardi syndrome in a male infant. *Journal of Pediatrics*, **1980**, 96, 286-7	3.6	35
364	The Q121/Q121 genotype of ENPP1/PC-1 is associated with lower BMI in non-diabetic whites. *Obesity*, **2007**, 15, 1-4	8	34

363	Relationship between CARD15, SLC22A4/5, and DLG5 polymorphisms and early-onset inflammatory bowel diseases: an Italian multicentric study. *Inflammatory Bowel Diseases*, **2006**, 12, 355-61	4.5	34
362	Auxological evaluation in patients with DiGeorge/velocardiofacial syndrome (deletion 22q11.2 syndrome). *Genetics in Medicine*, **2001**, 3, 30-3	8.1	34
361	Analysis of the elastin gene in 60 patients with clinical diagnosis of Williams syndrome. *Human Genetics*, **1995**, 96, 444-8	6.3	34
360	Duplication of Glu37 in the switch I region of HRAS impairs effector/GAP binding and underlies Costello syndrome by promoting enhanced growth factor-dependent MAPK and AKT activation. *Human Molecular Genetics*, **2010**, 19, 790-802	5.6	33
359	Noonan syndrome and aortic coarctation **1998**, 80, 160-162		33
358	Atypical phenotypes and clinical variability in a large Italian family with DYT1-primary torsion dystonia. *Movement Disorders*, **2006**, 21, 1782-4	7	33
357	Structural, Functional, and Clinical Characterization of a Novel PTPN11 Mutation Cluster Underlying Noonan Syndrome. *Human Mutation*, **2017**, 38, 451-459	4.7	32
356	Olfactory dysfunction in Parkinsonism caused by PINK1 mutations. *Movement Disorders*, **2009**, 24, 2350-7	7	32
355	Costello syndrome: clinical diagnosis in the first year of life. *European Journal of Pediatrics*, **2008**, 167, 621-8	4.1	32
354	Clinical phenotype and molecular characterization of 6q terminal deletion syndrome: Five new cases. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1944-9	2.5	32
353	Variations in the NMDA receptor subunit 2B gene (GRIN2B) and schizophrenia: a case-control study. *American Journal of Medical Genetics Part A*, **2004**, 128B, 27-9		32
352	Specific congenital heart defects in RSH/Smith-Lemli-Opitz syndrome: postulated involvement of the sonic hedgehog pathway in syndromes with postaxial polydactyly or heterotaxia. *Birth Defects Research Part A: Clinical and Molecular Teratology*, **2003**, 67, 149-53		32
351	Novel EXT1 and EXT2 mutations identified by DHPLC in Italian patients with multiple osteochondromas. *Human Mutation*, **2005**, 26, 280	4.7	32
350	Association study of a promoter polymorphism of UFD1L gene with schizophrenia. *American Journal of Medical Genetics Part A*, **2001**, 105, 529-33		32
349	Male hypogonadism in myotonic dystrophy is related to (CTG)n triplet mutation. *Journal of Endocrinological Investigation*, **1994**, 17, 381-3	5.2	32
348	Rare copy number variants and congenital heart defects in the 22q11.2 deletion syndrome. *Human Genetics*, **2016**, 135, 273-85	6.3	31
347	Exome sequencing in a family with intellectual disability, early onset spasticity, and cerebellar atrophy detects a novel mutation in EXOSC3. *Neurogenetics*, **2013**, 14, 247-50	3	31
346	Search of somatic GATA4 and NKX2.5 gene mutations in sporadic septal heart defects. *European Journal of Medical Genetics*, **2011**, 54, 306-9	2.6	31

345	Expression of receptors for native and chemically modified low-density lipoproteins in brain microvessels. *FEBS Letters*, **1997**, 401, 53-8	3.8	31
344	Isolation and characterization of a novel gene from the DiGeorge chromosomal region that encodes for a mediator subunit. *Genomics*, **2001**, 74, 320-32	4.3	31
343	Aberrant Function of the C-Terminal Tail of HIST1H1E Accelerates Cellular Senescence and Causes Premature Aging. *American Journal of Human Genetics*, **2019**, 105, 493-508	11	30
342	3q29 Microdeletion: a mental retardation disorder unassociated with a recognizable phenotype in two mother-daughter pairs. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 1777-81	2.5	30
341	KBG syndrome. *Orphanet Journal of Rare Diseases*, **2006**, 1, 50	4.2	30
340	Mutations of UFD1L are not responsible for the majority of cases of DiGeorge Syndrome/velocardiofacial syndrome without deletions within chromosome 22q11. *American Journal of Human Genetics*, **1999**, 65, 247-9	11	30
339	Prenatal diagnosis of recurrence of cystic hygroma with normal chromosomes. *Prenatal Diagnosis*, **1984**, 4, 383-6	3.2	30
338	Microbiome Analytics of the Gut Microbiota in Patients With Juvenile Idiopathic Arthritis: A Longitudinal Observational Cohort Study. *Arthritis and Rheumatology*, **2019**, 71, 1000-1010	9.5	30
337	De novo mutations of the gene encoding the histone acetyltransferase KAT6B in two patients with Say-Barber/Biesecker/Young-Simpson syndrome. *American Journal of Medical Genetics, Part A*, **2013** , 161A, 884-8	2.5	29
336	CARD15 genotyping in inflammatory bowel disease patients by multiplex pyrosequencing. *Clinical Chemistry*, **2003**, 49, 1675-9	5.5	29
335	KBG syndrome in a cohort of Italian patients. *American Journal of Medical Genetics Part A*, **2004**, 131, 144-9		29
334	Dopamine D4 receptor (DRD4) polymorphism and adaptability trait during infancy: a longitudinal study in 1- to 5-month-old neonates. *Neurogenetics*, **2001**, 3, 79-82	3	29
333	Genomic structure, promoter characterisation and mutational analysis of the S100A7 gene: exclusion of a candidate for familial psoriasis susceptibility. *Human Genetics*, **1999**, 104, 130-4	6.3	29
332	-related intellectual disability syndrome: a recognisable entity. *Journal of Medical Genetics*, **2017**, 54, 613-623	5.8	28
331	Ebstein anomaly: Genetic heterogeneity and association with microdeletions 1p36 and 8p23.1. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 2196-202	2.5	28
330	High frequency of COH1 intragenic deletions and duplications detected by MLPA in patients with Cohen syndrome. *European Journal of Human Genetics*, **2010**, 18, 1133-40	5.3	28
329	Syndromic craniosynostosis due to complex chromosome 5 rearrangement and MSX2 gene triplication. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 2937-43	2.5	28
328	Familial recurrence of heart defects in subjects with congenitally corrected transposition of the great arteries. *American Journal of Medical Genetics, Part A*, **2005**, 137, 176-80	2.5	28

327	Congenital heart defects in recurrent reciprocal 1q21.1 deletion and duplication syndromes: rare association with pulmonary valve stenosis. *European Journal of Medical Genetics*, **2013**, 56, 144-9	2.6	27
326	A variant in the carboxyl-terminus of connexin 40 alters GAP junctions and increases risk for tetralogy of Fallot. *European Journal of Human Genetics*, **2013**, 21, 69-75	5.3	27
325	Expression study of survival motor neuron gene in human fetal tissues. *Biochemical and Molecular Medicine*, **1997**, 61, 102-6		27
324	Nablus mask-like facial syndrome is caused by a microdeletion of 8q detected by array-based comparative genomic hybridization. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1267-73	2.5	27
323	2q31.2q32.3 deletion syndrome: report of an adult patient. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 706-12	2.5	26
322	Design of novel three-phase PCL/TZ-HA biomaterials for use in bone regeneration applications. *Journal of Materials Science: Materials in Medicine*, **2010**, 21, 2569-81	4.5	26
321	Antenatal presentation of the oculo-auriculo-vertebral spectrum (OAVS). *American Journal of Medical Genetics, Part A*, **2006**, 140, 1573-9	2.5	26
320	Novel NF1 gene mutation in a Japanese patient with neurofibromatosis type 1 and a gastrointestinal stromal tumor. *Journal of Gastroenterology*, **2006**, 41, 378-82	6.9	26
319	PTPN11 gene mutations: linking the Gln510Glu mutation to the "LEOPARD syndrome phenotype". *European Journal of Pediatrics*, **2006**, 165, 803-5	4.1	26
318	Recurrent triploidy of maternal origin. *European Journal of Human Genetics*, **2003**, 11, 972-4	5.3	26
317	Single atrium, atrioventricular canal/postaxial hexodactyly indicating Ellis-van Creveld syndrome. *Human Genetics*, **1995**, 96, 251-3	6.3	26
316	Functional analysis of splicing mutations in exon 7 of NF1 gene. *BMC Medical Genetics*, **2007**, 8, 4	2.1	25
315	A single polymerase chain reaction-based protocol for detecting normal and expanded alleles in myotonic dystrophy. *Diagnostic Molecular Pathology*, **1998**, 7, 135-7		25
314	Detection by capillary electrophoresis of restriction fragment length polymorphism. Analysis of a polymerase chain reaction-amplified product of the DXS 164 locus in the dystrophin gene. *Journal of Chromatography A*, **1993**, 638, 277-81	4.5	25
313	Increased rate of superoxide ion generation in Fanconi anemia erythrocytes. *Biochemical and Biophysical Research Communications*, **1985**, 130, 127-32	3.4	25
312	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. *American Journal of Human Genetics*, **2020**, 107, 499-513	11	25
311	Nectin-4 mutations causing ectodermal dysplasia with syndactyly perturb the rac1 pathway and the kinetics of adherens junction formation. *Journal of Investigative Dermatology*, **2014**, 134, 2146-2153	4.3	24
310	Expanding CEP290 mutational spectrum in ciliopathies. *American Journal of Medical Genetics, Part A* , **2009**, 149A, 2173-80	2.5	24

309	Shells and heart: are human laterality and chirality of snails controlled by the same maternal genes?. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 2419-25	2.5	24
308	Complex rearrangement of chromosomes 7q21.13-q22.1 confirms the ectrodactyly-deafness locus and suggests new candidate genes. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 238-44	2.5	24
307	Contribution of IBD5 locus to clinical features of IBD patients. *American Journal of Gastroenterology* , **2006**, 101, 318-25	0.7	24
306	Autosomal recessive form of whistling face syndrome in sibs. *American Journal of Medical Genetics Part A*, **1989**, 33, 542-4		24
305	Complete Sequence of the 22q11.2 Allele in 1,053 Subjects with 22q11.2 Deletion Syndrome Reveals Modifiers of Conotruncal Heart Defects. *American Journal of Human Genetics*, **2020**, 106, 26-40	11	24
304	Deletion 2p15-16.1 syndrome: case report and review. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 2473-8	2.5	23
303	Analysis of the DAZ gene family in cryptorchidism and idiopathic male infertility. *Fertility and Sterility*, **2004**, 81, 1013-8	4.8	23
302	Absent pulmonary valve with intact ventricular septum and patent ductus arteriosus: a specific cardiac phenotype associated with deletion 18q syndrome. *American Journal of Medical Genetics, Part A*, **2005**, 138A, 185-6	2.5	23
301	T cell receptor repertoire and function in patients with DiGeorge syndrome and velocardiofacial syndrome. *Clinical and Experimental Immunology*, **2000**, 121, 127-32	6.2	23
300	Human elongation factor EF-1 beta: cloning and characterization of the EF1 beta 5a gene and assignment of EF-1 beta isoforms to chromosomes 2,5,15 and X. *Biochemical and Biophysical Research Communications*, **1993**, 197, 154-62	3.4	23
299	Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. *Clinical Epigenetics*, **2020**, 12, 7	7.7	23
298	Molecular Diversity and Associated Phenotypic Spectrum of Germline CBL Mutations. *Human Mutation*, **2015**, 36, 787-96	4.7	22
297	De-novo 2.15 Mb terminal Xq duplication involving MECP2 but not L1CAM gene in a male patient with mental retardation. *Clinical Dysmorphology*, **2009**, 18, 9-12	0.9	22
296	Familial deletions of chromosome 22q11 **1997**, 73, 95-96		22
295	Late onset sporadic Parkinson's disease caused by PINK1 mutations: clinical and functional study. *Movement Disorders*, **2008**, 23, 881-5	7	22
294	CRELD1 and GATA4 gene analysis in patients with nonsyndromic atrioventricular canal defects. *American Journal of Medical Genetics, Part A*, **2005**, 139, 236-8	2.5	22
293	Heterotaxy with left atrial isomerism in a patient with deletion 18p. *American Journal of Medical Genetics Part A*, **2000**, 94, 198-200		22
292	Baller-Gerold syndrome: case report and clinical and radiological review. *American Journal of Medical Genetics Part A*, **1992**, 42, 365-8		22

291	"Omic" investigations of protozoa and worms for a deeper understanding of the human gut "parasitome". *PLoS Neglected Tropical Diseases*, **2017**, 11, e0005916	4.8	22
290	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. *American Journal of Human Genetics*, **2018**, 103, 305-316	11	21
289	3p25.3 microdeletion of GABA transporters SLC6A1 and SLC6A11 results in intellectual disability, epilepsy and stereotypic behavior. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 3061-8	2.5	21
288	The policy of public health genomics in Italy. *Health Policy*, **2013**, 110, 214-9	3.2	21
287	Meta-omic platforms to assist in the understanding of NAFLD gut microbiota alterations: tools and applications. *International Journal of Molecular Sciences*, **2014**, 15, 684-711	6.3	21
286	Interaction of DIO2 T92A and PPARgamma2 P12A polymorphisms in the modulation of metabolic syndrome. *Obesity*, **2007**, 15, 2889-95	8	21
285	Novel Italian family supports clinical and genetic heterogeneity of primary adult-onset torsion dystonia. *Movement Disorders*, **2002**, 17, 392-7	7	21
284	Localization of a gene for familial patella aplasia-hypoplasia (PTLAH) to chromosome 17q21-22. *American Journal of Human Genetics*, **1999**, 65, 441-7	11	21
283	SERCA1 protein expression in muscle of patients with Brody disease and Brody syndrome and in cultured human muscle fibers. *Molecular Genetics and Metabolism*, **2013**, 110, 162-9	3.7	20
282	High specificity of quantitative methylation-specific PCR analysis for MGMT promoter hypermethylation detection in gliomas. *Journal of Biomedicine and Biotechnology*, **2009**, 2009, 531692		20
281	16p subtelomeric duplication: a clinically recognizable syndrome. *European Journal of Human Genetics*, **2009**, 17, 1135-40	5.3	20
280	TBX2 gene duplication associated with complex heart defect and skeletal malformations. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 2061-6	2.5	20
279	Structure and expression of the human ubiquitin fusion-degradation gene (UFD1L). *Biochimica Et Biophysica Acta Gene Regulatory Mechanisms*, **1998**, 1396, 158-62		20
278	Advances in the search for psoriasis susceptibility genes. *Molecular Genetics and Metabolism*, **2000**, 71, 250-5	3.7	20
277	Expression analysis and protein localization of the human HPC-1/syntaxin 1A, a gene deleted in Williams syndrome. *Genomics*, **1999**, 62, 525-8	4.3	20
276	A 45,X male with molecular evidence of a translocation of Y euchromatin onto chromosome 1. *Human Genetics*, **1990**, 86, 94-8	6.3	20
275	A serine-to-arginine (AGT-to-CGT) mutation in codon 549 of the CFTR gene in an Italian patient with severe cystic fibrosis. *Genomics*, **1991**, 9, 788-9	4.3	20
274	Overt cleft palate phenotype and TBX1 genotype correlations in velo-cardio-facial/DiGeorge/22q11.2 deletion syndrome patients. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2781-7	2.5	19

273	Atrioventricular canal defect in patients with RASopathies. *European Journal of Human Genetics*, **2013**, 21, 200-4	5.3	19
272	Three novel mutations in the ANK membrane protein cause craniometaphyseal dysplasia with variable conductive hearing loss. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 870-4	2.5	19
271	Apolipoprotein E genotypes in hospitalized elderly patients with vascular dementia. *Dementia and Geriatric Cognitive Disorders*, **2007**, 23, 327-33	2.6	19
270	Genetic testing in Italy, year 2004. *European Journal of Human Genetics*, **2006**, 14, 911-6	5.3	19
269	Disomy of distal Xq in males: case report and overview. *American Journal of Medical Genetics Part A*, **2004**, 128A, 165-9		19
268	A single-nucleotide polymorphism in the human bone morphogenetic protein-4 (BMP 4) gene. *Journal of Human Genetics*, **1999**, 44, 76-7	4.3	19
267	Discordant clinical outcome in type III spinal muscular atrophy sibships showing the same deletion pattern. *Neuromuscular Disorders*, **1996**, 6, 261-4	2.9	19
266	A de novo proximal 3q29 chromosome microduplication in a patient with oculo auriculo vertebral spectrum. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 797-801	2.5	18
265	Quantification of small non-coding RNAs allows an accurate comparison of miRNA expression profiles. *Journal of Biomedicine and Biotechnology*, **2009**, 2009, 659028		18
264	Tricuspid atresia and 22q11 deletion. *American Journal of Medical Genetics Part A*, **1997**, 72, 40-2		18
263	Clinical and molecular characterization of Italian patients affected by Cohen syndrome. *Journal of Human Genetics*, **2007**, 52, 1011-1017	4.3	18
262	Frequency and phenotypes of LRRK2 G2019S mutation in Italian patients with Parkinson's disease. *Movement Disorders*, **2006**, 21, 1232-5	7	18
261	Combined test for UGT1A1 -3279T-->G and A(TA)nTAA polymorphisms best predicts Gilbert's syndrome in Italian pediatric patients. *Genetic Testing and Molecular Biomarkers*, **2006**, 10, 121-5		18
260	Ventricular septal defect and deletion of chromosome 22q11: anatomical types and aortic arch anomalies. *European Journal of Pediatrics*, **2002**, 161, 116-7	4.1	18
259	Ext-mutation analysis in Italian sporadic and hereditary osteochondromas. *International Journal of Cancer*, **2001**, 95, 378-83	7.5	18
258	Expanding the phenotypic spectrum of truncating POGZ mutations: Association with CNS malformations, skeletal abnormalities, and distinctive facial dysmorphism. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1965-1969	2.5	17
257	Long-term survival and phenotypic spectrum in heterotaxy syndrome: A 25-year follow-up experience. *International Journal of Cardiology*, **2018**, 268, 100-105	3.2	17
256	Low-rate repetitive nerve stimulation protocol in an Italian cohort of patients affected by recessive myotonia congenita. *Journal of Clinical Neurophysiology*, **2011**, 28, 39-44	2.2	17

255	FGF17, a gene involved in cerebellar development, is downregulated in a patient with Dandy-Walker malformation carrying a de novo 8p deletion. *Neurogenetics*, **2011**, 12, 241-5	3	17
254	GIGYF2 variants are not associated with Parkinson's disease in Italy. *Movement Disorders*, **2009**, 24, 1867-8; author reply 1868-9	7	17
253	Clinical utility gene card for: Mayer-Rokitansky-Küster-Hauser syndrome. *European Journal of Human Genetics*, **2012**, 20,	5.3	17
252	Atrioventricular canal defect and postaxial polydactyly indicating phenotypic overlap of Ellis-van Creveld and Kaufman-McKusick syndromes. *Pediatric Cardiology*, **1997**, 18, 74-5	2.1	17
251	Association of syndromic mental retardation with an Xq12q13.1 duplication encompassing the oligophrenin 1 gene. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1718-24	2.5	17
250	Isolation and characterization of a novel transcript embedded within HIRA, a gene deleted in DiGeorge syndrome. *Molecular Genetics and Metabolism*, **1999**, 67, 227-35	3.7	17
249	Discordant clinical outcome in myotonic dystrophy relatives showing (CTG)n > 700 repeats. *Neuromuscular Disorders*, **1995**, 5, 157-9	2.9	17
248	Haplotype analysis to determine the position of a mutation among closely linked DNA markers. *Human Molecular Genetics*, **1993**, 2, 1007-14	5.6	17
247	Two pedigrees of autosomal dominant atrioventricular canal defect (AVCD): exclusion from the critical region on 8p. *American Journal of Medical Genetics Part A*, **1995**, 57, 483-8		17
246	Linkage disequilibrium for DNA haplotypes near the cystic fibrosis locus in two south European populations. *Human Genetics*, **1989**, 83, 175-8	6.3	17
245	Clinical spectrum of Kabuki-like syndrome caused by HNRNPK haploinsufficiency. *Clinical Genetics*, **2018**, 93, 401-407	4	16
244	Clinical utility gene card for: Joubert syndrome--update 2013. *European Journal of Human Genetics*, **2013**, 21,	5.3	16
243	Association analysis of GRIN2B, encoding N-methyl-D-aspartate receptor 2B subunit, and Alzheimer's disease. *Dementia and Geriatric Cognitive Disorders*, **2008**, 25, 287-92	2.6	16
242	Multiplex ligation-dependent probe amplification assay for simultaneous detection of Parkinson's disease gene rearrangements. *Movement Disorders*, **2007**, 22, 2274-8	7	16
241	Pyrosequencing for detection of mutations in the connexin 26 (GJB2) and mitochondrial 12S RNA (MTRNR1) genes associated with hereditary hearing loss. *Human Mutation*, **2002**, 20, 312-20	4.7	16
240	Mutational analysis of parkin gene by denaturing high-performance liquid chromatography (DHPLC) in essential tremor. *Parkinsonism and Related Disorders*, **2004**, 10, 357-62	3.6	16
239	Rapid scanning of myotubularin (MTM1) gene by denaturing high-performance liquid chromatography (DHPLC). *Neuromuscular Disorders*, **2002**, 12, 501-5	2.9	16
238	A new method for direct analysis of polymerase chain reaction-amplified human papillomavirus using DNA enzyme immunoassay. *International Journal of Clinical and Laboratory Research*, **1994**, 24, 223-6		16

237	Atrioventricular canal and 3C (cranio-cerebello-cardiac) syndrome. *American Journal of Medical Genetics Part A*, **1995**, 58, 97-8		16
236	Atrioventricular canal associated with trisomy 9. *Chest*, **1989**, 96, 1420-1	5.3	16
235	Dandy-Walker malformation and Wisconsin syndrome: novel cases add further insight into the genotype-phenotype correlations of 3q23q25 deletions. *Orphanet Journal of Rare Diseases*, **2013**, 8, 75	4.2	15
234	Molecular analysis of PRKAG2, LAMP2, and NKX2-5 genes in a cohort of 125 patients with accessory atrioventricular connection. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 1574-7	2.5	15
233	Compound heterozygosity for GDF5 in Du Pan type chondrodysplasia. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2116-21	2.5	15
232	DHPLC screening of ATM gene in Italian patients affected by ataxia-telangiectasia: fourteen novel ATM mutations. *Disease Markers*, **2006**, 22, 257-64	3.2	15
231	Autosomal dominant Brody disease cosegregates with a chromosomal (2;7)(p11.2;p12.1) translocation in an Italian family. *European Journal of Human Genetics*, **2004**, 12, 579-83	5.3	15
230	Ablepharon-macrostomia syndrome in a 46-year-old woman. *American Journal of Medical Genetics Part A*, **2004**, 127A, 96-8		15
229	Gene transfection efficiency of tracheal epithelial cells by DC-chol-DOPE/DNA complexes. *Biochimica Et Biophysica Acta - Biomembranes*, **1999**, 1419, 186-94	3.8	15
228	Analysis of apoB, HLADQ alpha, and D1S80 polymorphisms in the Italian population using the polymerase chain reaction. *American Journal of Human Biology*, **1992**, 4, 381-386	2.7	15
227	Genome-Wide Association Study to Find Modifiers for Tetralogy of Fallot in the 22q11.2 Deletion Syndrome Identifies Variants in the Locus on 5q14.3. *Circulation: Cardiovascular Genetics*, **2017**, 10,		14
226	Parent-of-Origin Effects in 15q11.2 BP1-BP2 Microdeletion (Burnside-Butler) Syndrome. *International Journal of Molecular Sciences*, **2019**, 20,	6.3	14
225	Understanding probioticsNrole in allergic children: the clue of gut microbiota profiling. *Current Opinion in Allergy and Clinical Immunology*, **2015**, 15, 495-503	3.3	14
224	Glutamine to arginine substitution at amino acid 84 of mammalian tribbles homolog TRIB3 and CKD in whites with type 2 diabetes. *American Journal of Kidney Diseases*, **2007**, 50, 688-9	7.4	14
223	Atrioventricular canal defect in Bardet-Biedl syndrome: clinical evidence supporting the link between atrioventricular canal defect and polydactyly syndromes with ciliary dysfunction536. *Genetics in Medicine*, **2006**, 8, 536-8	8.1	14
222	Nonsyndromic pulmonary valve stenosis and the PTPN11 gene. *American Journal of Medical Genetics Part A*, **2003**, 116A, 389-90		14
221	Association study between CAG trinucleotide repeats in the PCQAP gene (PC2 glutamine/Q-rich-associated protein) and schizophrenia. *American Journal of Medical Genetics Part A*, **2003**, 116B, 32-5		14
220	Fetal translocation between chromosomes 2, 18, and 21 resolved by fish. *Prenatal Diagnosis*, **1995**, 15, 278-81	3.2	14

219	Complex translocation t(9;21)(9;22)(q12p13)(q12q11) in the family of a child with 9p trisomy syndrome. *Human Genetics*, **1976**, 33, 73-6	6.3	14
218	Percentiles of serum uric acid and cardiometabolic abnormalities in obese Italian children and adolescents. *Italian Journal of Pediatrics*, **2017**, 43, 3	3.2	13
217	Gingival overgrowth, congenital generalized hypertrichosis, mental retardation and epilepsy: case report and overview. *Clinical Dysmorphology*, **2009**, 18, 205-8	0.9	13
216	The atrioventricular canal defect is the congenital heart disease connecting short rib-polydactyly and oral-facial-digital syndromes. *American Journal of Medical Genetics Part A*, **1997**, 68, 110-2		13
215	Congenital heart defect in sibs with discordant karyotypes. *American Journal of Medical Genetics Part A*, **1998**, 80, 169-72		13
214	Microarray analysis identifies a common set of cellular genes modulated by different HCV replicon clones. *BMC Genomics*, **2008**, 9, 309	4.5	13
213	Ten novel mutations in the human neurofibromatosis type 1 (NF1) gene in Italian patients. *Human Mutation*, **2002**, 20, 74-5	4.7	13
212	Familial recurrence of nonsyndromic congenital heart defects in first degree relatives of patients with deletion 22q11.2. *American Journal of Medical Genetics, Part A*, **2005**, 134A, 158-64	2.5	13
211	Reproductive history of a healthy woman with mosaic duplication of chromosome 4p. *Prenatal Diagnosis*, **2005**, 25, 283-5	3.2	13
210	UFD1L and CDC45L: a role in DiGeorge syndrome and related phenotypes?. *Trends in Genetics*, **1999** , 15, 251-4	8.5	13
209	Reduction of the DM-associated homeo domain protein (DMAHP) mRNA in different brain areas of myotonic dystrophy patients. *Neuromuscular Disorders*, **1999**, 9, 215-9	2.9	13
208	The mendelian basis of congenital heart defects. *Cardiology in the Young*, **1996**, 6, 264-271	1	13
207	Search for 22q11 deletion in non-syndromic conotruncal cardiac defects. *European Journal of Pediatrics*, **1996**, 155, 619-20	4.1	13
206	Exclusion of linkage with chromosome 21 in families with recurrence of non-Down atrioventricular canal. *Human Genetics*, **1994**, 94, 708-10	6.3	13
205	Potential of multiomics technology in precision medicine. *Current Opinion in Gastroenterology*, **2019** , 35, 491-498	3	13
204	Kinematic and diffusion tensor imaging definition of familial Marcus Gunn jaw-winking synkinesis. *PLoS ONE*, **2012**, 7, e51749	3.7	12
203	The difficult nosology of blepharophimosis-mental retardation syndromes: report on two siblings. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 459-65	2.5	12
202	Founder effects for ATM gene mutations in Italian Ataxia Telangiectasia families. *Annals of Human Genetics*, **2009**, 73, 532-9	2.2	12

201	Clinical lumping and molecular splitting of LEOPARD and NF1/NF1-Noonan syndromes. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1009-11	2.5	12
200	Autosomal dominant inheritance of aplasia cutis congenita and congenital heart defect: a possible link to the Adams-Oliver syndrome. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2842-4	2.5	12
199	Analysis of CARD15 gene variants in Italian pediatric patients with inflammatory bowel diseases. *Journal of Pediatrics*, **2005**, 147, 272-3	3.6	12
198	ZFPM2/FOG2 and HEY2 genes analysis in nonsyndromic tricuspid atresia. *American Journal of Medical Genetics, Part A*, **2005**, 133A, 68-70	2.5	12
197	Diagnosis of DiGeorge and Williams syndromes using FISH analysis of peripheral blood smears. *Molecular and Cellular Probes*, **1999**, 13, 303-7	3.3	12
196	Postzygotic instability of the myotonic dystrophy p[AGC] in repeat supported by larger expansions in muscle and reduced amplifications in sperm. *Journal of Neurology*, **1995**, 242, 379-83	5.5	12
195	Red blood cell adenine nucleotides abnormalities in Down syndrome. *American Journal of Medical Genetics Part A*, **1985**, 20, 131-5		12
194	Absence of chromosome heterogeneity between classical Fanconi anemia and the Estren-Dameshek type. *Cancer Genetics and Cytogenetics*, **1980**, 2, 349-360		12
193	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. *American Journal of Human Genetics*, **2021**, 108, 502-516	11	12
192	Infantile cortical hyperostosis and COL1A1 mutation in four generations. *European Journal of Pediatrics*, **2011**, 170, 1385-90	4.1	11
191	Cutis laxa in Kabuki make-up syndrome. *Journal of the American Academy of Dermatology*, **2005**, 53, S247-51	4.5	11
190	Familial aggregation of genetically heterogeneous hypertrophic cardiomyopathy: a boy with LEOPARD syndrome due to PTPN11 mutation and his nonsyndromic father lacking PTPN11 mutations. *Birth Defects Research Part A: Clinical and Molecular Teratology*, **2004**, 70, 95-8		11
189	Familial recurrence of nonsyndromic interrupted aortic arch and truncus arteriosus with atrioventricular canal. *Teratology*, **2000**, 61, 329-31		11
188	The link between cytogenetics and mendelism. *Biomedicine and Pharmacotherapy*, **1995**, 49, 83-93	7.5	11
187	First-trimester prenatal diagnosis of spinal muscular atrophy using microsatellite markers. *Prenatal Diagnosis*, **1994**, 14, 459-62	3.2	11
186	Di George anomaly with atrioventricular canal. *Chest*, **1991**, 99, 242-3	5.3	11
185	Polymorphic DNA haplotypes and delta F508 deletion in 212 Italian CF families. *Human Genetics*, **1990**, 85, 420-1	6.3	11
184	Expanding the clinical spectrum associated with PACS2 mutations. *Clinical Genetics*, **2019**, 95, 525-531	4	10

183	KBG syndrome: Common and uncommon clinical features based on 31 new patients. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1073-1083	2.5	10
182	Delineating the psychiatric and behavioral phenotype of recurrent 2q13 deletions and duplications. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2018**, 177, 397-405	3.5	10
181	Heterozygous missense mutations in NFATC1 are associated with atrioventricular septal defect. *Human Mutation*, **2018**, 39, 1428-1441	4.7	10
180	Clinical and Neurobehavioral Features of Three Novel Kabuki Syndrome Patients with Mosaic KMT2D Mutations and a Review of Literature. *International Journal of Molecular Sciences*, **2017**, 19,	6.3	10
179	Cardiovascular malformations in Adams-Oliver syndrome. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 1175-7	2.5	10
178	Atrioventricular canal defect as a sign of laterality defect in Ellis-van Creveld and polydactyly syndromes with ciliary and Hedgehog signaling dysfunction. *Pediatric Cardiology*, **2012**, 33, 874-5	2.1	10
177	Germline mosaicism in neurofibromatosis type 1 due to a paternally derived multi-exon deletion. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1467-73	2.5	10
176	Sequence-specific modification of a beta-thalassemia locus by small DNA fragments in human erythroid progenitor cells. *Haematologica*, **2007**, 92, 129-30	6.6	10
175	Duplication 18q21.31-q22.2. *American Journal of Medical Genetics, Part A*, **2007**, 143, 343-8	2.5	10
174	Pigmented villonodular synovitis in a patient with Noonan syndrome and SOS1 gene mutation. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2966-7	2.5	10
173	Apolipoprotein E genotypes in mild cognitive impairment subtypes. *Journal of the American Geriatrics Society*, **2006**, 54, 1965-6	5.6	10
172	Confirmation of Nablus mask-like facial syndrome. *American Journal of Medical Genetics Part A*, **2003**, 121A, 283-5		10
171	A novel family with an unusual early-onset generalized dystonia. *Movement Disorders*, **2005**, 20, 81-6	7	10
170	Molecular characterization of the H319Q galactosemia mutation. *Human Molecular Genetics*, **1993**, 2, 325-6	5.6	10
169	3Ncreatine kinase (M-type) polymorphisms linked to myotonic dystrophy in Italian and Spanish populations. *Human Genetics*, **1991**, 87, 654-6	6.3	10
168	Genetic differences in cystic fibrosis patients with and without pancreatic insufficiency. An Italian collaborative study. *Human Genetics*, **1990**, 84, 435-8	6.3	10
167	Monitoring Perinatal Gut Microbiota in Mouse Models by Mass Spectrometry Approaches: Parental Genetic Background and Breastfeeding Effects. *Frontiers in Microbiology*, **2016**, 7, 1523	5.7	10
166	Duplications of GPC3 and GPC4 genes in symptomatic female carriers of Simpson-Golabi-Behmel syndrome type 1. *European Journal of Medical Genetics*, **2019**, 62, 243-247	2.6	9

165	Azoospermia in a man with a constitutional ring 22 chromosome. *European Journal of Medical Genetics*, **2010**, 53, 389-91	2.6	9
164	Expanding the phenotype of duplication of the Rubinstein-Taybi region on 16p13.3. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 2867-70	2.5	9
163	Bilateral frontoparietal polymicrogyria (BFPP) syndrome secondary to a 16q12.1-q21 chromosome deletion involving GPR56 gene. *Clinical Genetics*, **2009**, 76, 573-6	4	9
162	Conotruncal heart defects and chromosome 22q11 microdeletion. *Journal of Pediatrics*, **1997**, 130, 675-7	3.6	9
161	Genetic testing for paediatric neurological disorders. *Lancet Neurology, The*, **2008**, 7, 1113-26	24.1	9
160	Congenital intrahepatic portosystemic venous shunt: an unusual feature in LEOPARD syndrome and in neurofibromatosis type 1. *American Journal of Medical Genetics, Part A*, **2005**, 134, 457-8	2.5	9
159	Individual haploinsufficient loci and the complex phenotype of DiGeorge syndrome. *Trends in Molecular Medicine*, **2000**, 6, 10-1		9
158	Cloning and characterization of the gene encoding human NPL4, a protein interacting with the ubiquitin fusion-degradation protein (UFD1L). *Gene*, **2001**, 275, 39-46	3.8	9
157	Simultaneous detection of delta F508, G542X, N1303K, G551D, and 1717-1G-->A cystic fibrosis alleles by a multiplex DNA enzyme immunoassay. *International Journal of Clinical and Laboratory Research*, **1995**, 25, 142-5		9
156	The prevalence of HPV16DNA in normal and pathological cervical scrapes using the polymerase chain reaction. *Gynecologic Oncology*, **1992**, 46, 33-6	4.9	9
155	Deletion 2q31.3----2q33.3: gene dosage effect of ribulose 5-phosphate 3-epimerase. *Human Genetics*, **1988**, 79, 92	6.3	9
154	Tetraploidy (92,XXYY) in an acute nonlymphocytic leukemia (M1) patient following autologous bone marrow transplantation. *Cancer Genetics and Cytogenetics*, **1988**, 36, 69-75		9
153	Ultrastructural and biological characterization of human choroid cell cultures transformed by Simian Virus 40. *In Vitro*, **1983**, 19, 443-52		9
152	JAG1 mutation in a patient with deletion 22q11.2 syndrome and tetralogy of Fallot. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 3133-6	2.5	8
151	Nablus mask-like facial syndrome: deletion of chromosome 8q22.1 is necessary but not sufficient to cause the phenotype. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2091-9	2.5	8
150	Genetic dosage compensation in a family with velo-cardio-facial/DiGeorge/22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 548-54	2.5	8
149	Combinatorial sequencing-by-hybridization: analysis of the NF1 gene. *Genetic Testing and Molecular Biomarkers*, **2006**, 10, 8-17		8
148	Concordant familial segregation of atrial septal defect and Axenfeld-Rieger anomaly in father and son. *Clinical Dysmorphology*, **2006**, 15, 203-206	0.9	8

147	Identification of type A, B, E, and F botulinum neurotoxin genes and of botulinum neurotoxigenic clostridia by denaturing high-performance liquid chromatography. *Applied and Environmental Microbiology*, **2004**, 70, 4170-6	4.8	8
146	A new observation of acro-cardio-facial syndrome substantiates interindividual clinical variability. *American Journal of Medical Genetics, Part A*, **2005**, 136, 84-6	2.5	8
145	Hyperthrophic cardiomyopathy and the PTPN11 gene. *American Journal of Medical Genetics, Part A*, **2005**, 136, 93-4	2.5	8
144	Mapping of a new autosomal dominant nonsyndromic hearing loss locus (DFNA30) to chromosome 15q25-26. *European Journal of Human Genetics*, **2001**, 9, 667-71	5.3	8
143	De novo deletions of the 5q13 region and prenatal diagnosis of spinal muscular atrophy. *Prenatal Diagnosis*, **1995**, 15, 93-4	3.2	8
142	Identification of six novel CFTR mutations in a sample of Italian cystic fibrosis patients. *Molecular and Cellular Probes*, **1995**, 9, 135-7	3.3	8
141	Diagnosis of DiGeorge syndrome in nuclei released from archival autoptic heart specimens using fluorescence in situ hybridization. *Human Genetics*, **1996**, 97, 414-7	6.3	8
140	Delta F508 gene deletion and prenatal diagnosis of cystic fibrosis in Italian and Spanish families. *Prenatal Diagnosis*, **1990**, 10, 413-4	3.2	8
139	SCUBE3 loss-of-function causes a recognizable recessive developmental disorder due to defective bone morphogenetic protein signaling. *American Journal of Human Genetics*, **2021**, 108, 115-133	11	8
138	Reassessment of the 12q15 deletion syndrome critical region. *European Journal of Medical Genetics*, **2017**, 60, 220-223	2.6	7
137	Connexin 26 variant carriers have a better gastrointestinal health: is this the heterozygote advantage?. *European Journal of Human Genetics*, **2015**, 23, 563-564	5.3	7
136	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome or Fowler syndrome: Report of a family and insight into the diseases mechanism. *Molecular Genetics & Genomic Medicine*, **2018**, 6, 446-451	2.3	7
135	Can modern biology interpret the mystery of the birth of Christ?. *Journal of Maternal-Fetal and Neonatal Medicine*, **2015**, 28, 240-4	2	7
134	Hypoplastic left heart syndrome and 21q22.3 deletion. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 579-86	2.5	7
133	Hypoplastic left heart syndrome in patients with Kabuki syndrome. *Pediatric Cardiology*, **2010**, 31, 1111-3	2.1	7
132	Noonan syndrome with cardiac left-sided obstructive lesions. *Human Genetics*, **1997**, 99, 289	6.3	7
131	Severe, obstructive biventricular hypertrophy in a patient with Costello syndrome: Clinical impact and management. *International Journal of Cardiology*, **2008**, 130, e108-10	3.2	7
130	Reassessment of holoprosencephaly-diencephalic hamartoblastoma (HDH) association. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 277-84	2.5	7

129	Autosomal dominant transmission of nonsyndromic diastasis recti and weakness of the linea alba. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 254-6	2.5	7
128	Genetics of congenital heart diseases in syndromic and non-syndromic patients: new advances and clinical implications. *Journal of Cardiovascular Medicine*, **2007**, 8, 7-11	1.9	7
127	Diffuse coronary dilation in a young patient with LEOPARD syndrome. *International Journal of Cardiology*, **2006**, 112, e35-7	3.2	7
126	Denaturing HPLC-based assay for molecular screening of nondeletional mutations causing alpha-thalassemias. *Clinical Chemistry*, **2004**, 50, 1242-5	5.5	7
125	Validation of dHPLC for molecular diagnosis of beta-thalassemia in Southern Italy. *Genetic Testing and Molecular Biomarkers*, **2003**, 7, 269-75		7
124	A single strand conformation polymorphism-based carrier test for spinal muscular atrophy. *Genetic Testing and Molecular Biomarkers*, **2001**, 5, 33-7		7
123	A novel mutation (R271X) in the myotubularin gene causes a severe miotubular myopathy. *Human Heredity*, **1999**, 49, 59-60	1.1	7
122	Atrioventricular canal defect and hypoplastic left heart syndrome as discordant congenital heart defects in twins. *Teratology*, **1999**, 60, 206-8		7
121	Linkage analysis of Fanconi anaemia in Italy and mapping of the complementation group A gene. *Human Genetics*, **1997**, 99, 93-7	6.3	7
120	Three new mutations (P183T, V150L, 528insG) and eleven sequence polymorphisms in Italian patients with galactose-1-phosphate uridyltransferase (GALT) deficiency. *Human Mutation*, **1996**, 8, 369-72	4.7	7
119	Regional mapping of hexokinase-1 within the short arm of chromosome 10. *Human Heredity*, **1984**, 34, 156-60	1.1	7
118	Increased activity of glutathione S-transferase and fast decay of reduced glutathione in Fanconi anemia erythrocytes. *Acta Haematologica*, **1984**, 71, 143-4	2.7	7
117	Multiplex Ligation-Dependent Probe Amplification Analysis ofGATA4Gene Copy Number Variations in Patients with Isolated Congenital Heart Disease. *Disease Markers*, **2010**, 28, 287-292	3.2	7
116	Confirmation of BRD4 haploinsufficiency role in Cornelia de Lange-like phenotype and delineation of a 19p13.12p13.11 gene contiguous syndrome. *Annals of Human Genetics*, **2019**, 83, 100-109	2.2	7
115	An additional patient with a homozygous mutation in DCPS contributes to the delination of Al-Raqad syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2781-2786	2.5	7
114	Small 4p16.3 deletions: Three additional patients and review of the literature. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2501-2508	2.5	7
113	Atypical cardiac defects in patients with RASopathies: Updated data on CARNET study. *Birth Defects Research*, **2020**, 112, 725-731	2.9	6
112	Dysmorphologic assessment in 115 Mayer-Rokitansky-Küster-Hauser patients. *Clinical Dysmorphology*, **2015**, 24, 95-101	0.9	6

111	Ablepharon macrostomia syndrome: A distinct genetic entity clinically related to the group of FRAS-FREM complex disorders. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 3012-7	2.5	6
110	Atrioventricular canal defect and associated genetic disorders: new insights into polydactyly syndromes. *Neurology International*, **2011**, 1, 7	0	6
109	Familial spinal neurofibromatosis due to a multiexonic NF1 gene deletion. *Neurogenetics*, **2011**, 12, 233-40	3	6
108	A nationwide genetic testing survey in Italy, year 2007. *Genetic Testing and Molecular Biomarkers*, **2010**, 14, 17-22	1.6	6
107	A 6-year-old child with Fryns syndrome: further delineation of the natural history of the condition in survivors. *European Journal of Medical Genetics*, **2009**, 52, 421-5	2.6	6
106	Response to local dihydrotestosterone treatment in a patient with partial androgen-insensitivity syndrome due to a novel mutation in the androgen receptor gene. *American Journal of Medical Genetics Part A*, **2002**, 107, 259-60		6
105	Inosine-containing primers in human papillomavirus detection by polymerase chain reaction. *Biomedicine and Pharmacotherapy*, **1992**, 46, 167-9	7.5	6
104	Regional mapping of the locus for hexokinase-1 (HK1). *Human Genetics*, **1982**, 62, 181	6.3	6
103	TARP syndrome: Long-term survival, anatomic patterns of congenital heart defects, differential diagnosis and pathogenetic considerations. *European Journal of Medical Genetics*, **2019**, 62, 103534	2.6	6
102	A heterozygous, intragenic deletion of CNOT2 recapitulates the phenotype of 12q15 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1615-1621	2.5	5
101	Data on cardiac defects, morbidity and mortality in patients affected by RASopathies. CARNET study results. *Data in Brief*, **2018**, 16, 649-654	1.2	5
100	SOS1 mutations in Noonan syndrome: Cardiomyopathies and not only congenital heart defects! Report of six patients including two novel variants and literature review. *American Journal of Medical Genetics, Part A*, **2019**, 179, 2083-2090	2.5	5
99	CHARGE syndrome due to deletion of region upstream of CHD7 gene START codon. *BMC Medical Genetics*, **2015**, 16, 78	2.1	5
98	Behavioral phenotype in Costello syndrome with atypical mutation: a case report. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2015**, 168B, 66-71	3.5	5
97	Association of DiGeorge anomaly and caudal dysplasia sequence in a neonate born to a diabetic mother. *Cardiology in the Young*, **2013**, 23, 14-7	1	5
96	Phenocopy of Wolf-Hirschhorn syndrome in a patient with duplication 12q13.3q14.1. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 546-8	2.5	5
95	Association of apolipoprotein E and angiotensin converting enzyme gene polymorphisms with the multidimensional impairment in older patients. *Rejuvenation Research*, **2009**, 12, 239-47	2.6	5
94	Deletion 22q11 and isolated congenital heart disease. *International Journal of Cardiology*, **2008**, 123, 364-5	3.2	5

93	A novel patient with Cooks syndrome supports splitting from "classic" brachydactyly type B. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 195-9	2.5	5
92	Five cases of supernumerary small ring chromosomes 1: heterogeneity and genotype-phenotype correlation. *European Journal of Medical Genetics*, **2007**, 50, 94-102	2.6	5
91	Primary hypothyroidism and osteopenia associated with Neuhauser syndrome. *American Journal of Medical Genetics Part A*, **2002**, 111, 412-4		5
90	Functional characterization of the 5Nflanking region of human ubiquitin fusion degradation 1 like gene (UFD1L). *Cell Biochemistry and Function*, **2002**, 20, 163-70	4.2	5
89	Fine mapping of a distinctive autosomal dominant vacuolar neuromyopathy using 11 novel microsatellite markers from chromosome band 19p13.3. *European Journal of Human Genetics*, **2000**, 8, 809-12	5.3	5
88	Identification of multiple transcribed sequences from the spinal muscular atrophy region of human chromosome 5. *Biochemical and Biophysical Research Communications*, **1995**, 206, 294-301	3.4	5
87	Detection of eight beta-thalassemia mutations using a DNA enzyme immunoassay. *International Journal of Clinical and Laboratory Research*, **1996**, 26, 136-9		5
86	Deletion analysis of the simple tandem repeat loci physically linked to the spinal muscular atrophy locus. *Human Mutation*, **1996**, 7, 198-201	4.7	5
85	A tool for the molecular analysis of an early lethal disease: slide-PCR in spinal muscular atrophy patients. *Molecular and Cellular Probes*, **1993**, 7, 221-6	3.3	5
84	Plasmid DNA and low-frequency electromagnetic fields. *Biomedicine and Pharmacotherapy*, **1993**, 47, 101-5	7.5	5
83	North Eurasian origin of the myotonic dystrophy mutation. *Human Mutation*, **1994**, 4, 79-81	4.7	5
82	Assignment of the slow troponin T (TNNT1) gene to chromosome 19 using polymerase chain reaction. *Human Genetics*, **1992**, 88, 697-8	6.3	5
81	Identification of 4 ataxia telangiectasia cell lines hypersensitive to gamma-irradiation but not to hydrogen peroxide. *Mutation Research DNA Repair*, **1989**, 218, 143-8		5
80	Association of Bright Liver With the PNPLA3 I148M Gene Variant in 1-Year-Old Toddlers. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 2163-2170	5.6	4
79	Progeroid syndrome with characteristic facial appearance and hand anomalies in father and son. *American Journal of Medical Genetics Part A*, **1997**, 73, 227-9		4
78	Familial recurrence of transposition of the great arteries. *Lancet, The*, **1998**, 351, 1661	40	4
77	Reticulate vascular lesions and a large head. *Pediatric Dermatology*, **2007**, 24, 555-6	1.9	4
76	Abnormal neuronal migration defect in the severe variant subtype of Adams-Oliver syndrome. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1622-3	2.5	4

75	Detection of a rare beta-globin nonsense mutation [codon 59 (AAG-->TAG)] in an Italian family. *Hemoglobin*, **2006**, 30, 405-7	0.6	4
74	Ellis-van Creveld Syndrome with hydrometrocolpos is not linked to chromosome arm 4p or 20p. *American Journal of Medical Genetics Part A*, **2004**, 126A, 319-23		4
73	Different expression of the myotonin protein kinase gene in discrete areas of human brain. *Biochemical and Biophysical Research Communications*, **1995**, 216, 489-94	3.4	4
72	The dynamic genomics of myotonic dystrophy and its clinical relevance: an overview. *Biomedicine and Pharmacotherapy*, **1993**, 47, 321-30	7.5	4
71	TurnerÑ syndrome with atrioventricular canal. *Pediatric Cardiology*, **1991**, 12, 245-6	2.1	4
70	Increased erythrocyte adenosine deaminase activity without haemolytic anaemia. *Human Heredity*, **1986**, 36, 37-40	1.1	4
69	Acute lymphocytic and myelomonocytic leukemia associated with low platelet counts and a 21q- marker chromosome. *Human Genetics*, **1981**, 57, 329-31	6.3	4
68	Severe, early onset hypertrophic cardiomyopathy in a family with LEOPARD syndrome. *Journal of Prenatal Medicine*, **2008**, 2, 24-6		4
67	Deficiency of MFSD7c results in microcephaly-associated vasculopathy in Fowler syndrome. *Journal of Clinical Investigation*, **2020**, 130, 4081-4093	15.9	4
66	Skeletal abnormalities are common features in Aym□Gripp syndrome. *Clinical Genetics*, **2020**, 97, 362-369	4	4
65	Cross-correlation of virome-bacteriome-host-metabolome to study respiratory health. *Trends in Microbiology*, **2021**,	12.4	4
64	Unclassifiable pattern of hypopigmentation in a patient with mosaic partial 12p tetrasomy without Pallister-Killian syndrome. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1943-1946	2.5	3
63	Co-occurrence of mutations in KIF7 and KIAA0556 in Joubert syndrome with ocular coloboma, pituitary malformation and growth hormone deficiency: a case report and literature review. *BMC Pediatrics*, **2020**, 20, 120	2.6	3
62	The splice c.1815G>A variant in KIAA0586 results in a phenotype bridging short-rib-polydactyly and oral-facial-digital syndrome: A case report and literature review. *Medicine (United States)*, **2020**, 99, e19169	1.8	3
61	Hypopigmented skin patches in 17q21.31 microdeletion syndrome: expanding the spectrum of cutaneous findings. *Clinical Dysmorphology*, **2014**, 23, 32-34	0.9	3
60	Acro-cardio-facial syndrome. *Orphanet Journal of Rare Diseases*, **2010**, 5, 25	4.2	3
59	Exclusion of Cx43 gene mutation as a major cause of criss-cross heart anomaly in man. *International Journal of Cardiology*, **2010**, 144, 300-2	3.2	3
58	X chromosome monosomy restricted to the left ventricle is not a major cause of isolated hypoplastic left heart. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1967-72	2.5	3

57	Familial recurrence of transposition of the great arteries and intact ventricular septum **1997**, 73, 93-94		3
56	Genomic instability associated with myotonic dystrophy does not involve p53 expression and activity. *Cell Biochemistry and Function*, **1998**, 16, 117-22	4.2	3
55	Hypochondrogenesis. *Pediatric Radiology*, **2006**, 36, 460-1	2.8	3
54	Additional evidence that PTPN11 mutations play only a minor role in the pathogenesis of non-syndromic atrioventricular canal defect. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1970-2	2.5	3
53	22q11 deletion syndrome: a review of some developmental biology aspects of the cardiovascular system. *Journal of Cardiovascular Medicine*, **2006**, 7, 77-85	1.9	3
52	Report of a third family with Oliver syndrome. *American Journal of Medical Genetics, Part A*, **2005**, 139A, 159-61	2.5	3
51	Confirmation of the mandibulofacial dysostosis, Toriello type. *American Journal of Medical Genetics Part A*, **1993**, 45, 534-5		3
50	Study of the effects on DNA of electromagnetic fields using clamped homogeneous electric field gel electrophoresis. *Biomedicine and Pharmacotherapy*, **1991**, 45, 451-4	7.5	3
49	1p13.2 deletion displays clinical features overlapping Noonan syndrome, likely related to NRAS gene haploinsufficiency. *Genetics and Molecular Biology*, **2016**, 39, 349-57	2	3
48	Sprengel anomaly in deletion 22q11.2 (DiGeorge/Velo-Cardio-Facial) syndrome. *American Journal of Medical Genetics, Part A*, **2016**, 170, 661-4	2.5	3
47	Relationship between glucose homeostasis and obesity in early life-A study of Italian children and adolescents. *Human Molecular Genetics*, **2021**,	5.6	3
46	Familial aggregation of "apple peel" intestinal atresia and cardiac left-sided obstructive lesions: A possible causal relationship with NOTCH1 gene mutations. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1570-1574	2.5	2
45	Cost-effectiveness of exome sequencing: an Italian pilot study on undiagnosed patients. *New Genetics and Society*, **2019**, 38, 249-263	1.9	2
44	First case of nonalcoholic steatohepatitis in a child with del(1p36) and dup (Xp22): review of the literature. *Clinical Dysmorphology*, **2018**, 27, 42-45	0.9	2
43	Survey of medical genetic services in Italy: year 2011. *BMC Health Services Research*, **2016**, 16, 96	2.9	2
42	The Challenge of Prenatal Diagnostic Work-Up of Maternally Inherited X-Linked Opitz G/BBB: Case Report and Literature Review. *Case Reports in Obstetrics and Gynecology*, **2015**, 2015, 830108	0.8	2
41	Analysis of intracellular distribution and apoptosis involvement of the Ufd1l gene product by over-expression studies. *Cell Biochemistry and Function*, **2003**, 21, 263-7	4.2	2
40	Association of deletion 22 and trisomy 21: a likely random association in patients with conotruncal heart defects. *American Journal of Medical Genetics, Part A*, **2005**, 134A, 1-2	2.5	2

39	The up-to-date molecular genetics of cystic fibrosis. *Biomedicine and Pharmacotherapy*, **1994**, 48, 455-63	7.5	2
38	Cytogenetics of Mendelian mutations associated with cancer proneness. *Cancer Genetics and Cytogenetics*, **1987**, 26, 85-94		2
37	Alkaline phosphatase expression in human chorionic villi. *Pathobiology*, **1987**, 55, 34-41	3.6	2
36	SPRED2 loss-of-function causes a recessive Noonan syndrome-like phenotype. *American Journal of Human Genetics*, **2021**, 108, 2112-2129	11	2
35	Delayed appearance of 3-methylglutaconic aciduria in neonates with early onset metabolic cardiomyopathies: A potential pitfall for the diagnosis. *American Journal of Medical Genetics, Part A*, **2020**, 182, 64-70	2.5	2
34	PPP1R21-related syndromic intellectual disability: Report of an adult patient and review. *American Journal of Medical Genetics, Part A*, **2020**, 182, 3014-3022	2.5	2
33	Copy number variation analysis implicates novel pathways in patients with oculo-auriculo-vertebral-spectrum and congenital heart defects. *Clinical Genetics*, **2021**, 100, 268-279	4	2
32	Deletion 8p syndrome **1998**, 75, 534		2
31	Interstitial 10q21.1q23.31 Duplication due to Meiotic Recombination of a Paternal Balanced Complex Rearrangement: Cytogenetic and Molecular Characterization. *Cytogenetic and Genome Research*, **2017**, 151, 179-185	1.9	1
30	Left ventricular non compaction with aortic valve anomalies: A recurrent feature of 22q11.2 distal deletion syndrome. *European Journal of Medical Genetics*, **2015**, 58, 406-8	2.6	1
29	AuthorsN response to J Thomas: "If not parthenogenesis why not in vivo embryogenesisN with Mary as a birth mother". *Journal of Maternal-Fetal and Neonatal Medicine*, **2015**, 28, 1917	2	1
28	Fused Omics Data Models Reveal Gut Microbiome Signatures Specific of Inactive Stage of Juvenile Idiopathic Arthritis in Pediatric Patients. *Microorganisms*, **2020**, 8,	4.9	1
27	RDDR: a dysmorphology diagnostic network for newborns in central Italy. *Journal of Maternal-Fetal and Neonatal Medicine*, **2012**, 25 Suppl 4, 121-3	2	1
26	A triploid fetus further expands etiological heterogeneity in holoprosencephaly-diencephalic hamartoblastoma. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1391-3	2.5	1
25	A novel locus for autosomal dominant cone and cone-rod dystrophies maps to the 6p gene cluster of retinal dystrophies. *Investigative Ophthalmology and Visual Science*, **2005**, 46, 3539-44		1
24	Nasal cartilage aplasia in a family with facioaudiosymphalangism syndrome. *American Journal of Otolaryngology - Head and Neck Medicine and Surgery*, **2000**, 21, 405-8	2.8	1
23	Three novel mutations causing a truncated protein within the RP2 gene in Italian families with X-linked retinitis pigmentosa. *Mutation Research - Mutation Research Genomics*, **2001**, 432, 79-82		1
22	Discrete subaortic stenosis. *American Heart Journal*, **1994**, 127, 1665-6	4.9	1

21	PCR amplification and silver stain detection of genomic DNA fragments. *Trends in Genetics*, **1989**, 5, 293	8.5	1
20	The Q121/Q121 Genotype of ENPP1/PC-1 Is Associated with Lower BMI in Non-diabetic Whites **2007**, 15, 1		1
19	Genetics of Ebstein Anomaly **2014**, 25-30		1
18	Genome-Wide DNA Methylation Analysis of a Cohort of 41 Patients Affected by Oculo-Auriculo-Vertebral Spectrum (OAVS). *International Journal of Molecular Sciences*, **2021**, 22,	6.3	1
17	First Report of Low-Rate Mosaicism for 20q11.21q12 Deletion and Delineation of the Associated Disorder. *Cytogenetic and Genome Research*, **2018**,	1.9	1
16	Apparent third patient with cutaneous mastocytosis, microcephaly, conductive hearing loss, and microtia. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 2270-3	2.5	0
15	Functional Genomics for Undiagnosed Patients: The Impact of Small GTPases Signaling Dysregulation at Pan-Embryo Developmental Scale. *Frontiers in Cell and Developmental Biology*, **2021**, 9, 642235	5.7	0
14	Clinical Application of Easychip 8x15K Platform in 4106 Pregnancies Without Ultrasound Anomalies. *Reproductive Sciences*, **2021**, 28, 1142-1149	3	0
13	Homozygous and Gene Variants in a Boy with Growth Hormone Deficiency and Early Onset Osteoporosis. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	0
12	Complex Presentation of Hao-Fountain Syndrome Solved by Exome Sequencing Highlighting Co-Occurring Genomic Variants. *Genes*, **2022**, 13, 889	4.2	0
11	A familial chromosomal complex rearrangement confirms RUNX1T1 as a causative gene for intellectual disability and suggests that 1p22.1p21.3 duplication is likely benign. *Molecular Cytogenetics*, **2019**, 12, 26	2	
10	Screening for 22q11.2 microdeletion in adults with tetralogy of Fallot. *Heart*, **2011**, 97, 860; author reply 860	5.1	
9	Confirmation of microcephaly-facio-cardio-skeletal Hadziselimovic type syndrome. *Clinical Dysmorphology*, **2009**, 18, 135-138	0.9	
8	Obituary of Dr. Angelo di George. *Italian Journal of Pediatrics*, **2010**, 36, 22	3.2	
7	Atrioventricular Canal Defect: Anatomical and Genetic Characteristics **2007**, 244-247		
6	Familial Recurrence Risks of Congenital Heart Defects **2007**, 232-234		
5	Comment on the paper of Qumsiyeh et al. (Am J Med Genet 41: 99-101, 1991). *American Journal of Medical Genetics Part A*, **1992**, 44, 844-6		
4	Reply to letter by M. Poot and H. Hoehn. *Human Genetics*, **1990**, 84, 582	6.3	

3	DOWN SYNDROME AND THYROID FUNCTION. *Lancet, The*, **1976**, 307, 362	40
2	Application of the Capillary DNA Chromatography in the Paternity Testing Using APOB Amplified Alleles. *Advances in Forensic Haemogenetics*, **1994**, 136-138	
1	Diagnosis of DiGeorge syndrome in nuclei released from archival autoptic heart specimens using fluorescence in situ hybridization. *Human Genetics*, **1996**, 97, 414-417	6.3