Willy Lissens

List of Publications by Year
in descending order

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124 papers	6,026 citations	100601 38 h-index	90395 73 g-index
125 all docs	125 docs citations	125 times ranked	5540 citing authors

#	Article	IF	Citations
1	Clinical implementation of gene panel testing for lysosomal storage diseases. Molecular Genetics & Genomic Medicine, 2019, 7, e00527.	0.6	18
2	Bi-allelic variants inCOL3A1encoding the ligand to GPR56 are associated with cobblestone-like cortical malformation, white matter changes and cerebellar cysts. Journal of Medical Genetics, 2017, 54, 432-440.	1.5	34
3	Sertoli Cell-Only Syndrome: Behind the Genetic Scenes. BioMed Research International, 2016, 2016, 1-7.	0.9	22
4	Convert your favorite protein modeling program into a mutation predictor: â€œMODICTâ€• BMC Bioinformatics, 2016, 17, 425.	1.2	2
5	SCN4A variants and Brugada syndrome: phenotypic and genotypic overlap between cardiac and skeletal muscle sodium channelopathies. European Journal of Human Genetics, 2016, 24, 400-407.	1.4	33
6	I-PV: a CIRCOS module for interactive protein sequence visualization. Bioinformatics, 2016, 32, 447-449.	1.8	6
7	Antithrombin heparin binding site deficiency: A challenging diagnosis of a not so benign thrombophilia. Thrombosis Research, 2015, 135, 1179-1185.	0.8	28
8	Analysis of the whole mitochondrial genome: translation of the Ion Torrent Personal Genome Machine system to the diagnostic bench?. European Journal of Human Genetics, 2015, 23, 41-48.	1.4	33
9	Genetic causes of male infertility. Annales D'Endocrinologie, 2014, 75, 109-111.	0.6	23
10	Clinical variability in neurohepatic syndrome due to combined mitochondrial DNA depletion and Gaucher disease. Molecular Genetics and Metabolism Reports, 2014, 1, 223-231.	0.4	2
11	A Bumpy Ride on the Diagnostic Bench of Massive Parallel Sequencing, the Case of the Mitochondrial Genome. PLoS ONE, 2014, 9, e112950.	1.1	13
12	Elaborating the phenotypic spectrum associated with mutations in ARFGEF2: Case study and literature review. European Journal of Paediatric Neurology, 2013, 17, 666-670.	0.7	12
13	Fluorescence imaging of mitochondria in cultured skin fibroblasts: a useful method for the detection of oxidative phosphorylation defects. Pediatric Research, 2012, 72, 232-240.	1.1	16
14	X chromosomal mutations and spermatogenic failure. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 1864-1872.	1.8	24
15	Reliable and Sensitive Detection of Fragile X (Expanded) Alleles in Clinical Prenatal DNA Samples with a Fast Turnaround Time. Journal of Molecular Diagnostics, 2012, 14, 560-568.	1.2	10
16	Genetic causes of spermatogenic failure. Asian Journal of Andrology, 2012, 14, 40-48.	0.8	168
17	Identification of two de novo mutations responsible for type I antithrombin deficiency. Thrombosis and Haemostasis, 2012, 107, 187-189.	1.8	4
18	Proteomic analysis in giant axonal neuropathy: New insights into disease mechanisms. Muscle and Nerve, 2012, 46, 246-256.	1.0	12

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19	Severe male factor: Genetic consequences and recommendations for genetic testing. , 2012, , 324-335.		0
20	A novel mutation in the SCN4A responsible for cold-induced myotonia with normal electromyography findings on room temperature. Journal of the Neurological Sciences, 2011, 308, 162-164.	0.3	1
21	Late onset painful cold-aggravated myotonia: Three families with SCN4A L1436P mutation. Neuromuscular Disorders, 2011, 21, 590-593.	0.3	12
22	Mutation analysis of three genes in patients with maturation arrest of spermatogenesis and couples with recurrent miscarriages. Reproductive BioMedicine Online, 2011, 22, 65-71.	1.1	31
23	DISCORDANCE FOR RETINITIS PIGMENTOSA IN TWO MONOZYGOTIC TWIN PAIRS. Retina, 2011, 31, 1164-1169.	1.0	5
24	Complex III staining in blue native polyacrylamide gels. Journal of Inherited Metabolic Disease, 2011, 34, 741-747.	1.7	21
25	What about gr/gr deletions and male infertility? Systematic review and meta-analysis. Human Reproduction Update, 2011, 17, 197-209.	5.2	82
26	Cancer predisposing missense and protein truncatingBARD1mutations in non-BRCA1orBRCA2breast cancer families. Human Mutation, 2010, 31, E1175-E1185.	1.1	86
27	Giant axonal neuropathy caused by compound heterozygosity for a maternally inherited microdeletion and a paternal mutation within the GAN gene. American Journal of Medical Genetics, Part A, 2010, 152A, 2802-2804.	0.7	19
28	Defining the Pathogenesis of the Human Atp12p W94R Mutation Using a Saccharomyces cerevisiae Yeast Model. Journal of Biological Chemistry, 2010, 285, 4099-4109.	1.6	17
29	Male infertility and the involvement of the X chromosome. Human Reproduction Update, 2009, 15, 623-637.	5.2	54
30	Subcomplexes of mitochondrial complex V reveal mutations in mitochondrial DNA. Electrophoresis, 2009, 30, 3565-3572.	1.3	28
31	Lactic Acidosis in a Newborn With Adrenal Calcifications. Pediatric Research, 2009, 66, 317-322.	1.1	5
32	Do we need to search for gr/gr deletions in infertile men in a clinical setting?. Human Reproduction, 2008, 23, 1193-1199.	0.4	32
33	Is there a role for the nuclear export factor 2 gene in male infertility?. Fertility and Sterility, 2008, 90, 1787-1791.	0.5	18
34	A New Missense Mutation in theCASRGene in Familial Interstitial Lung Disease with Hypocalciuric Hypercalcemia and Defective Granulocyte Function. American Journal of Respiratory and Critical Care Medicine, 2008, 177, 558-559.	2.5	8
35	Severe male factor. , 2008, , 343-356.		0
36	Two Novel Mitochondrial DNA Mutations in Muscle Tissue of a Patient With Limb-Girdle Myopathy. Archives of Neurology, 2007, 64, 1339.	4.9	12

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37	Molecular Analysis in Two Siblings African Patients with Severe Form of Hunter Syndrome: Identification of a Novel (p.Y54X) Nonsense Mutation. Journal of Tropical Pediatrics, 2007, 53, 434-437.	0.7	3
38	Sanfilippo Syndrome Type D. Archives of Neurology, 2007, 64, 1629.	4.9	29
39	A case of thyroid hormone resistance: Prospective follow-up during pregnancy and obstetric outcome. European Journal of Internal Medicine, 2007, 18, 253-254.	1.0	9
40	A single mutation in the GALC gene is responsible for the majority of late onset Krabbe disease patients in the Catania (Sicily, Italy) region. Human Mutation, 2007, 28, 742-742.	1.1	38
41	A new family with the mitochondrial tRNAGLU gene mutation m.14709T>C presenting with hydrops fetalis. European Journal of Paediatric Neurology, 2007, 11, 17-20.	0.7	15
42	Serine Protease Activity and Residual LEKTI Expression Determine Phenotype in Netherton Syndrome. Journal of Investigative Dermatology, 2006, 126, 1609-1621.	0.3	163
43	Preimplantation genetic diagnosis for Marfan syndrome. Fertility and Sterility, 2006, 86, 310-320.	0.5	40
44	A Novel Mitochondrial Transfer RNAAsn Mutation Causing Multiorgan Failure. Archives of Neurology, 2006, 63, 1194.	4.9	29
45	Alterations of the USP26 gene in Caucasian men. Journal of Developmental and Physical Disabilities, 2006, 29, 614-617.	3.6	32
46	Gas chromatographicâ€“mass spectrometric analysis of N-acetylated amino acids: The first case of aminoacylase I deficiency. Analytica Chimica Acta, 2006, 571, 191-199.	2.6	26
47	Diagnostic Value of Immunostaining in Cultured Skin Fibroblasts from Patients with Oxidative Phosphorylation Defects. Pediatric Research, 2006, 59, 2-6.	1.1	20
48	Disorders of Pyruvate Metabolism and the Tricarboxylic Acid Cycle. , 2006, , 161-174.		8
49	Possible role of USP26 in patients with severely impaired spermatogenesis. European Journal of Human Genetics, 2005, 13, 336-340.	1.4	85
50	A family with pyruvate dehydrogenase complex deficiency due to a novel C>T substitution at nucleotide position 407 in exon 4 of the X-linked Î•1Î± gene. European Journal of Pediatrics, 2005, 164, 99-103.	1.3	13
51	A novel L1CAM mutation with L1 spectrum disorders. Prenatal Diagnosis, 2005, 25, 57-59.	1.1	15
52	PGD for autosomal dominant polycystic kidney disease type 1. Molecular Human Reproduction, 2005, 11, 65-71.	1.3	43
53	The choice and outcome of the fertility treatment of 38 couples in whom the male partner has a Yq microdeletion. Human Reproduction, 2005, 20, 1887-1896.	0.4	65
54	Preimplantation genetic diagnosis for neurofibromatosis type 1. Molecular Human Reproduction, 2005, 11, 381-387.	1.3	55

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55	SYCP3 mutations are uncommon in patients with azoospermia. Fertility and Sterility, 2005, 84, 1019-1020.	0.5	35
56	Mutations c.459+1G>A and p.P426L in the ARSA gene: Prevalence in metachromatic leukodystrophy patients from European countries. Molecular Genetics and Metabolism, 2005, 86, 353-359.	0.5	27
57	Novel universal approach for preimplantation genetic diagnosis of Â-thalassaemia in combination with HLA matching of embryos. Human Reproduction, 2004, 19, 700-708.	0.4	98
58	Respiratory chain complex V deficiency due to a mutation in the assembly gene ATP12. Journal of Medical Genetics, 2004, 41, 120-124.	1.5	175
59	Expression pattern of the Y-linked PRY gene suggests a function in apoptosis but not in spermatogenesis. Molecular Human Reproduction, 2004, 10, 15-21.	1.3	39
60	Analysis of the mitochondrial encoded subunits of complex I in 20 patients with a complex I deficiency. European Journal of Paediatric Neurology, 2004, 8, 299-306.	0.7	6
61	Early onset Huntington disease: a neuronal degeneration syndrome. European Journal of Pediatrics, 2004, 163, 717-721.	1.3	48
62	Idiopathic non-obstructive azoospermia or severe oligozoospermia: a cross-sectional study in 61 Greek men. Journal of Developmental and Physical Disabilities, 2004, 27, 101-107.	3.6	7
63	Genetically heterogeneous selective intestinal malabsorption of vitamin B12: Founder effects, consanguinity, and high clinical awareness explain aggregations in Scandinavia and the Middle East. Human Mutation, 2004, 23, 327-333.	1.1	73
64	Intergenerational Instability of the Expanded CTG Repeat in the DMPK Gene: Studies in Human Gametes and Preimplantation Embryos. American Journal of Human Genetics, 2004, 75, 325-329.	2.6	69
65	Clinical and diagnostic characteristics of complex III deficiency due to mutations in theBCS1Lgene. , 2003, 121A, 126-131.		86
66	Improving clinical preimplantation genetic diagnosis for cystic fibrosis by duplex PCR using two polymorphic markers or one polymorphic marker in combination with the detection of the ÂF508 mutation. Molecular Human Reproduction, 2003, 9, 559-567.	1.3	34
67	Loss of DNA-dependent dimerization of the transcription factor SOX9 as a cause for campomelic dysplasia. Human Molecular Genetics, 2003, 12, 1439-1447.	1.4	122
68	Preimplantation genetic diagnosis for Charcot-Marie-Tooth disease type 1A. Molecular Human Reproduction, 2003, 9, 429-435.	1.3	31
69	Preimplantation genetic diagnosis for Huntington's disease with exclusion testing. European Journal of Human Genetics, 2002, 10, 591-598.	1.4	77
70	PGD in the lab for triplet repeat diseases ? myotonic dystrophy, Huntington's disease and Fragile-X syndrome. Molecular and Cellular Endocrinology, 2001, 183, S77-S85.	1.6	71
71	Preimplantation genetic diagnosis for spinal and bulbar muscular atrophy (SBMA). Human Genetics, 2001, 108, 494-498.	1.8	20
72	Imprinting analysis in spermatozoa prepared for intracytoplasmic sperm injection (ICSI). Journal of Developmental and Physical Disabilities, 2001, 24, 87-94.	3.6	31

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73	The role of USP9Y and DBY in infertile patients with severely impaired spermatogenesis. Molecular Human Reproduction, 2001, 7, 691-693.	1.3	13
74	DNA Methylation Analysis in Immature Testicular Sperm Cells at Different Developmental Stages. Urologia Internationalis, 2001, 67, 151-155.	0.6	20
75	Characterization of the genomic organization, localization and expression of four PRY genes (PRY1,) Tj ETQq1 1 0.784314 rgBT /Over	1.3	20
76	A New Mitochondrial Point Mutation in the Transfer RNALeu Gene in a Patient With a Clinical Phenotype Resembling Kearns-Sayre Syndrome. Archives of Neurology, 2001, 58, 1113.	4.9	46
77	Mutations in the X-linked pyruvate dehydrogenase (E1) ? subunit gene (PDHA1) in patients with a pyruvate dehydrogenase complex deficiency. Human Mutation, 2000, 15, 209-219.	1.1	191
78	Embryo implantation after biopsy of one or two cells from cleavage-stage embryos with a view to preimplantation genetic diagnosis. Prenatal Diagnosis, 2000, 20, 1030-1037.	1.1	120
79	Two pregnancies after preimplantation genetic diagnosis for osteogenesis imperfecta type I and type IV. Human Genetics, 2000, 106, 605-613.	1.8	8
80	Preimplantation genetic diagnosis for medium-chain acyl-CoA dehydrogenase (MCAD) deficiency. Molecular Human Reproduction, 2000, 6, 1165-1168.	1.3	13
81	Study of DNA-methylation patterns at chromosome 15q11-q13 in children born after ICSI reveals no imprinting defects. Molecular Human Reproduction, 2000, 6, 1049-1053.	1.3	97
82	Analysis of Exonic Mutations Leading to Exon Skipping in Patients with Pyruvate Dehydrogenase E1Î± Deficiency. Pediatric Research, 2000, 48, 748-753.	1.1	17
83	Validation of a simple Yq deletion screening programme in an ICSI candidate population. Molecular Human Reproduction, 2000, 6, 291-297.	1.3	51
84	The Brussels' experience of more than 5 years of clinical preimplantation genetic diagnosis. Human Reproduction Update, 2000, 6, 364-373.	5.2	54
85	Two pregnancies after preimplantation genetic diagnosis for osteogenesis imperfecta type I and type IV. Human Genetics, 2000, 106, 605-613.	1.8	16
86	Mutations in the X-linked pyruvate dehydrogenase (E1) Î± subunit gene (PDHA1) in patients with a pyruvate dehydrogenase complex deficiency. Human Mutation, 2000, 15, 209.	1.1	9
87	Fluorescent PCR and automated fragment analysis in preimplantation genetic diagnosis for 21-hydroxylase deficiency in congenital adrenal hyperplasia. Molecular Human Reproduction, 1999, 5, 691-696.	1.3	34
88	Molecular analysis of the cystic fibrosis gene reveals a high frequency of the intron 8 splice variant 5T in Egyptian males with congenital bilateral absence of the vas deferens. Molecular Human Reproduction, 1999, 5, 10-13.	1.3	34
89	Evaluation of Parental Mitochondrial Inheritance in Neonates Born after Intracytoplasmic Sperm Injection. American Journal of Human Genetics, 1999, 65, 463-473.	2.6	75
90	Pyruvate dehydrogenase complex deficiency and altered respiratory chain function in a patient with Kearnsâ€“Sayre/MELAS overlap syndrome and A3243G mtDNA mutation. Journal of the Neurological Sciences, 1998, 157, 206-213.	0.3	25

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91	Fluorescent PCR and automated fragment analysis for the clinical application of preimplantation genetic diagnosis of myotonic dystrophy (Steinert's disease). Molecular Human Reproduction, 1998, 4, 791-796.	1.3	101
92	Pregnancy after preimplantation genetic diagnosis for Charcot-Marie- Tooth disease type 1A. Molecular Human Reproduction, 1998, 4, 978-984.	1.3	44
93	8b The genetics of male infertility in relation to cystic fibrosis. Bailliere's Clinical Obstetrics and Gynaecology, 1997, 11, 797-817.	0.6	17
94	Biochemical and genetic studies of four patients with pyruvate dehydrogenase E1Î± deficiency. Human Genetics, 1997, 99, 785-792.	1.8	52
95	Pearson marrow pancreas syndrome: a molecular study and clinical management. Clinical Genetics, 1997, 51, 338-342.	1.0	37
96	Î²-Glucuronidase P408S, P415L mutations: evidence that both mutations combine to produce an MPS VII allele in certain Mexican patients. Human Genetics, 1996, 98, 281-284.	1.8	23
97	Mutation analysis of the pyruvate dehydrogenase E1Î± gene in eight patients with a pyruvate dehydrogenase complex deficiency. , 1996, 7, 46-51.		36
98	Cystic fibrosis, Duchenne muscular dystrophy and preimplantation genetic disorders. Human Reproduction Update, 1996, 2, 531-539.	5.2	8
99	Genetics: The use of epididymal and testicular spermatozoa for intracytoplasmic sperm injection: the genetic implications for male infertility. Human Reproduction, 1995, 10, 2031-2043.	0.4	230
100	Identification of two novel mutations in the cystic fibrosis gene: 1898 + 3Aâ†'C and 2711delT. Human Mutation, 1995, 6, 188-189.	1.1	0
101	Normal pregnancy after preimplantation DNA diagnosis of a dystrophin gene deletion. Prenatal Diagnosis, 1995, 15, 351-358.	1.1	64
102	Preimplantation Diagnosis of the Cystic Fibrosis Î"F508 Mutation: What of the Other Two Embryos?-Reply. JAMA - Journal of the American Medical Association, 1995, 274, 127.	3.8	0
103	Pyruvate dehydrogenase deficiency in a female due to a 4 base pair deletion in exon 10 of the E1Î± gene. Human Molecular Genetics, 1995, 4, 307-308.	1.4	13
104	Bilateral striatal necrosis with a novel point mutation in the mitochondrial ATPase 6 gene. Pediatric Neurology, 1995, 13, 242-246.	1.0	139
105	Nonsense mutation Arg197stop in a Dutch family with type 1 hereditary antithrombin (AT) deficiency causing thrombophilia. Thrombosis Research, 1995, 78, 251-254.	0.8	3
106	Mutations in the Cystic Fibrosis Gene in Patients with Congenital Absence of the Vas Deferens. New England Journal of Medicine, 1995, 332, 1475-1480.	13.9	959
107	Aberrant Splicing of Exon 6 in the Pyruvate Denydrogenase-ElÎ± mRNA Linked to a Silent Mutation in a Large Family with Leigh's Encephalomyelopathy. Pediatric Research, 1994, 36, 707-712.	1.1	37
108	Preimplantation diagnosis of genetic and chromosomal disorders. Journal of Assisted Reproduction and Genetics, 1994, 11, 236-243.	1.2	46

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109	Microsurgical epididymal sperm aspiration and intracytoplasmic sperm injection: a new effective approach to infertility as a result of congenital bilateral absence of the vas deferens. Fertility and Sterility, 1994, 61, 1045-1051.	0.5	327
110	Amplification of X-and Y-chromosome-specific regions from single human blastomeres by polymerase chain reaction for sexing of preimplantation embryos. Human Reproduction, 1994, 9, 716-720.	0.4	37
111	Detection of more than 94% cystic fibrosis mutations in a sample of belgian population and identification of four novel mutations. Human Mutation, 1993, 2, 16-20.	1.1	33
112	Molecular analysis of a patient with hydrops fetalis caused by Î²-glucuronidase deficiency, and evidence for additional pseudogenes. Human Mutation, 1993, 2, 443-445.	1.1	24
113	Polymerase chain reaction analysis of the cystic fibrosis Î”F508 mutation in human blastomeres following oocyte injection of a single sperm from a carrier. Prenatal Diagnosis, 1993, 13, 873-880.	1.1	32
114	Pyruvate dehydrogenase deficiency: Clinical and biochemical diagnosis. Pediatric Neurology, 1993, 9, 216-220.	1.0	63
115	El Pyruvate Dehydrogenase Deficiency in a Child with Motor Neuropathy. Pediatric Research, 1993, 33, 284-288.	1.1	40
116	Two novel mutations of the porphobilinogen deaminase gene in acute intermittent porphyria. Human Molecular Genetics, 1993, 2, 1735-1736.	1.4	30
117	Efficiency of polymerase chain reaction assay for cystic fibrosis in single human blastomeres according to the presence or absence of nuclei. Fertility and Sterility, 1993, 59, 815-819.	0.5	28
118	Efficiency and accuracy of polymerase-chain-reaction assay for cystic fibrosis allele Î”F508 in single cell. Lancet, The, 1992, 339, 1190-1192.	6.3	45
119	Î²-N-acetylhexosaminidase activity in human oocytes and preimplantation embryos. Human Reproduction, 1992, 7, 1278-1280.	0.4	6
120	Pyruvate dehydrogenase (PDH) deficiency caused by a 21-base pair insertion mutation in the ElÎ± subunit. Human Genetics, 1992, 88, 649-652.	1.8	24
121	The deletion F508 is the major gene mutation in a representative Belgian cystic fibrosis population. Human Genetics, 1990, 85, 395-396.	1.8	3
122	CarP, a novel gene regulating the transcription of the carbamoylphosphate synthetase operon of Escherichia coli. Journal of Molecular Biology, 1988, 204, 857-865.	2.0	26
123	In Vitro Synthesis of Escherichia coli Carbamoylphosphate Synthase: Evidence for Participation of the Arginine Repressor in Cumulative Repression. Journal of Bacteriology, 1980, 141, 58-66.	1.0	27
124	Genes and infertility. , 0, , 113-126.		0