Willy Lissens

List of Publications by Year
in descending order

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124	6,026	87888 38	79698 73
papers	citations	h-index	g-index

125	125	125	5164
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations in the Cystic Fibrosis Gene in Patients with Congenital Absence of the Vas Deferens. New England Journal of Medicine, 1995, 332, 1475-1480.	27.0	959
2	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.	1.0	327
3	Genetics: The use of epididymal and testicular spermatozoa for intracytoplasmic sperm injection: the genetic implications for male infertility. Human Reproduction, 1995, 10, 2031-2043.	0.9	230
4	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.	2.5	191
5	Respiratory chain complex V deficiency due to a mutation in the assembly gene ATP12. Journal of Medical Genetics, 2004, 41, 120-124.	3.2	175
6	Genetic causes of spermatogenic failure. Asian Journal of Andrology, 2012, 14, 40-48.	1.6	168
7	Serine Protease Activity and Residual LEKTI Expression Determine Phenotype in Netherton Syndrome. Journal of Investigative Dermatology, 2006, 126, 1609-1621.	0.7	163
8	Bilateral striatal necrosis with a novel point mutation in the mitochondrial ATPase 6 gene. Pediatric Neurology, 1995, 13, 242-246.	2.1	139
9	Loss of DNA-dependent dimerization of the transcription factor SOX9 as a cause for campomelic dysplasia. Human Molecular Genetics, 2003, 12, 1439-1447.	2.9	122
10	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.	2.3	120
11	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.	2.8	101
12	Novel universal approach for preimplantation genetic diagnosis of Â-thalassaemia in combination with HLA matching of embryos. Human Reproduction, 2004, 19, 700-708.	0.9	98
13	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.	2.8	97
14	Clinical and diagnostic characteristics of complex III deficiency due to mutations in theBCS1Lgene. , 2003, 121A, 126-131.		86
15	Cancer predisposing missense and protein truncatingBARD1mutations in non-BRCA1orBRCA2breast cancer families. Human Mutation, 2010, 31, E1175-E1185.	2.5	86
16	Possible role of USP26 in patients with severely impaired spermatogenesis. European Journal of Human Genetics, 2005, 13, 336-340.	2.8	85
17	What about gr/gr deletions and male infertility? Systematic review and meta-analysis. Human Reproduction Update, 2011, 17, 197-209.	10.8	82
18	Preimplantation genetic diagnosis for Huntington's disease with exclusion testing. European Journal of Human Genetics, 2002, 10, 591-598.	2.8	77

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19	Evaluation of Parental Mitochondrial Inheritance in Neonates Born after Intracytoplasmic Sperm Injection. American Journal of Human Genetics, 1999, 65, 463-473.	6.2	75
20	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.	2.5	73
21	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.	3.2	71
22	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.	6.2	69
23	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.	0.9	65
24	Normal pregnancy after preimplantation DNA diagnosis of a dystrophin gene deletion. Prenatal Diagnosis, 1995, 15, 351-358.	2.3	64
25	Pyruvate dehydrogenase deficiency: Clinical and biochemical diagnosis. Pediatric Neurology, 1993, 9, 216-220.	2.1	63
26	Preimplantation genetic diagnosis for neurofibromatosis type 1. Molecular Human Reproduction, 2005, 11, 381-387.	2.8	55
27	The Brussels' experience of more than 5 years of clinical preimplantation genetic diagnosis. Human Reproduction Update, 2000, 6, 364-373.	10.8	54
28	Male infertility and the involvement of the X chromosome. Human Reproduction Update, 2009, 15, 623-637.	10.8	54
29	Biochemical and genetic studies of four patients with pyruvate dehydrogenase E1Î± deficiency. Human Genetics, 1997, 99, 785-792.	3.8	52
30	Validation of a simple Yq deletion screening programme in an ICSI candidate population. Molecular Human Reproduction, 2000, 6, 291-297.	2.8	51
31	Early onset Huntington disease: a neuronal degeneration syndrome. European Journal of Pediatrics, 2004, 163, 717-721.	2.7	48
32	Preimplantation diagnosis of genetic and chromosomal disorders. Journal of Assisted Reproduction and Genetics, 1994, 11, 236-243.	2.5	46
33	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.	4.5	46
34	Efficiency and accuracy of polymerase-chain-reaction assay for cystic fibrosis allele Î”F508 in single cell. Lancet, The, 1992, 339, 1190-1192.	13.7	45
35	Pregnancy after preimplantation genetic diagnosis for Charcot-Marie- Tooth disease type 1A. Molecular Human Reproduction, 1998, 4, 978-984.	2.8	44
36	PGD for autosomal dominant polycystic kidney disease type 1. Molecular Human Reproduction, 2005, 11, 65-71.	2.8	43

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37	El Pyruvate Dehydrogenase Deficiency in a Child with Motor Neuropathy. Pediatric Research, 1993, 33, 284-288.	2.3	40
38	Preimplantation genetic diagnosis for Marfan syndrome. Fertility and Sterility, 2006, 86, 310-320.	1.0	40
39	Expression pattern of the Y-linked PRY gene suggests a function in apoptosis but not in spermatogenesis. Molecular Human Reproduction, 2004, 10, 15-21.	2.8	39
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.	2.5	38
41	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.	2.3	37
42	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.	0.9	37
43	Pearson marrow pancreas syndrome: a molecular study and clinical management. Clinical Genetics, 1997, 51, 338-342.	2.0	37
44	Mutation analysis of the pyruvate dehydrogenase E1Î± gene in eight patients with a pyruvate dehydrogenase complex deficiency. , 1996, 7, 46-51.		36
45	SYCP3 mutations are uncommon in patients with azoospermia. Fertility and Sterility, 2005, 84, 1019-1020.	1.0	35
46	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.	2.8	34
47	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.	2.8	34
48	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.	2.8	34
49	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.	3.2	34
50	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.	2.5	33
51	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.	2.8	33
52	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.	2.8	33
53	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.	2.3	32
54	Alterations of the USP26 gene in Caucasian men. Journal of Developmental and Physical Disabilities, 2006, 29, 614-617.	3.6	32

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55	Do we need to search for gr/gr deletions in infertile men in a clinical setting?. Human Reproduction, 2008, 23, 1193-1199.	0.9	32
56	Imprinting analysis in spermatozoa prepared for intracytoplasmic sperm injection (ICSI). Journal of Developmental and Physical Disabilities, 2001, 24, 87-94.	3.6	31
57	Preimplantation genetic diagnosis for Charcot-Marie-Tooth disease type 1A. Molecular Human Reproduction, 2003, 9, 429-435.	2.8	31
58	Mutation analysis of three genes in patients with maturation arrest of spermatogenesis and couples with recurrent miscarriages. Reproductive BioMedicine Online, 2011, 22, 65-71.	2.4	31
59	Two novel mutations of the porphobilinogen deaminase gene in acute intermittent porphyria. Human Molecular Genetics, 1993, 2, 1735-1736.	2.9	30
60	A Novel Mitochondrial Transfer RNAAsn Mutation Causing Multiorgan Failure. Archives of Neurology, 2006, 63, 1194.	4.5	29
61	Sanfilippo Syndrome Type D. Archives of Neurology, 2007, 64, 1629.	4.5	29
62	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.	1.0	28
63	Subcomplexes of mitochondrial complex V reveal mutations in mitochondrial DNA. Electrophoresis, 2009, 30, 3565-3572.	2.4	28
64	Antithrombin heparin binding site deficiency: A challenging diagnosis of a not so benign thrombophilia. Thrombosis Research, 2015, 135, 1179-1185.	1.7	28
65	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.	1.1	27
66	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.	2.2	27
67	CarP, a novel gene regulating the transcription of the carbamoylphosphate synthetase operon of Escherichia coli. Journal of Molecular Biology, 1988, 204, 857-865.	4.2	26
68	Gas chromatographicâ€“mass spectrometric analysis of N-acetylated amino acids: The first case of aminoacylase I deficiency. Analytica Chimica Acta, 2006, 571, 191-199.	5.4	26
69	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.	0.6	25
70	Pyruvate dehydrogenase (PDH) deficiency caused by a 21-base pair insertion mutation in the ElÎ± subunit. Human Genetics, 1992, 88, 649-652.	3.8	24
71	Molecular analysis of a patient with hydrops fetalis caused by Î²-glucuronidase deficiency, and evidence for additional pseudogenes. Human Mutation, 1993, 2, 443-445.	2.5	24
72	X chromosomal mutations and spermatogenic failure. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 1864-1872.	3.8	24

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73	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.	3.8	23
74	Genetic causes of male infertility. Annales D'Endocrinologie, 2014, 75, 109-111.	1.4	23
75	Sertoli Cell-Only Syndrome: Behind the Genetic Scenes. BioMed Research International, 2016, 2016, 1-7.	1.9	22
76	Complex III staining in blue native polyacrylamide gels. Journal of Inherited Metabolic Disease, 2011, 34, 741-747.	3.6	21
77	Preimplantation genetic diagnosis for spinal and bulbar muscular atrophy (SBMA). Human Genetics, 2001, 108, 494-498.	3.8	20
78	DNA Methylation Analysis in Immature Testicular Sperm Cells at Different Developmental Stages. Urologia Internationalis, 2001, 67, 151-155.	1.3	20
79	Characterization of the genomic organization, localization and expression of four PRY genes (PRY1,) Tj ETQq1 1 0.784314 rgBT /Over	2.8	20
80	Diagnostic Value of Immunostaining in Cultured Skin Fibroblasts from Patients with Oxidative Phosphorylation Defects. Pediatric Research, 2006, 59, 2-6.	2.3	20
81	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.	1.2	19
82	Is there a role for the nuclear export factor 2 gene in male infertility?. Fertility and Sterility, 2008, 90, 1787-1791.	1.0	18
83	Clinical implementation of gene panel testing for lysosomal storage diseases. Molecular Genetics & Genomic Medicine, 2019, 7, e00527.	1.2	18
84	8b The genetics of male infertility in relation to cystic fibrosis. Bailliere's Clinical Obstetrics and Gynaecology, 1997, 11, 797-817.	0.6	17
85	Analysis of Exonic Mutations Leading to Exon Skipping in Patients with Pyruvate Dehydrogenase E1Î± Deficiency. Pediatric Research, 2000, 48, 748-753.	2.3	17
86	Defining the Pathogenesis of the Human Atp12p W94R Mutation Using a Saccharomyces cerevisiae Yeast Model. Journal of Biological Chemistry, 2010, 285, 4099-4109.	3.4	17
87	Two pregnancies after preimplantation genetic diagnosis for osteogenesis imperfecta type I and type IV. Human Genetics, 2000, 106, 605-613.	3.8	16
88	Fluorescence imaging of mitochondria in cultured skin fibroblasts: a useful method for the detection of oxidative phosphorylation defects. Pediatric Research, 2012, 72, 232-240.	2.3	16
89	A novel L1CAM mutation with L1 spectrum disorders. Prenatal Diagnosis, 2005, 25, 57-59.	2.3	15
90	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.	1.6	15

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91	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.	2.9	13
92	Preimplantation genetic diagnosis for medium-chain acyl-CoA dehydrogenase (MCAD) deficiency. Molecular Human Reproduction, 2000, 6, 1165-1168.	2.8	13
93	The role of USP9Y and DBY in infertile patients with severely impaired spermatogenesis. Molecular Human Reproduction, 2001, 7, 691-693.	2.8	13
94	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.	2.7	13
95	A Bumpy Ride on the Diagnostic Bench of Massive Parallel Sequencing, the Case of the Mitochondrial Genome. PLoS ONE, 2014, 9, e112950.	2.5	13
96	Two Novel Mitochondrial DNA Mutations in Muscle Tissue of a Patient With Limb-Girdle Myopathy. Archives of Neurology, 2007, 64, 1339.	4.5	12
97	Late onset painful cold-aggravated myotonia: Three families with SCN4A L1436P mutation. Neuromuscular Disorders, 2011, 21, 590-593.	0.6	12
98	Proteomic analysis in giant axonal neuropathy: New insights into disease mechanisms. Muscle and Nerve, 2012, 46, 246-256.	2.2	12
99	Elaborating the phenotypic spectrum associated with mutations in ARFGEF2: Case study and literature review. European Journal of Paediatric Neurology, 2013, 17, 666-670.	1.6	12
100	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.	2.8	10
101	A case of thyroid hormone resistance: Prospective follow-up during pregnancy and obstetric outcome. European Journal of Internal Medicine, 2007, 18, 253-254.	2.2	9
102	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.	2.5	9
103	Cystic fibrosis, Duchenne muscular dystrophy and preimplantation genetic disorders. Human Reproduction Update, 1996, 2, 531-539.	10.8	8
104	Two pregnancies after preimplantation genetic diagnosis for osteogenesis imperfecta type I and type IV. Human Genetics, 2000, 106, 605-613.	3.8	8
105	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.	5.6	8
106	Disorders of Pyruvate Metabolism and the Tricarboxylic Acid Cycle. , 2006, , 161-174.		8
107	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
108	Î²-N-acetylhexosaminidase activity in human oocytes and preimplantation embryos. Human Reproduction, 1992, 7, 1278-1280.	0.9	6

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109	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.	1.6	6
110	I-PV: a CIRCOS module for interactive protein sequence visualization. Bioinformatics, 2016, 32, 447-449.	4.1	6
111	Lactic Acidosis in a Newborn With Adrenal Calcifications. Pediatric Research, 2009, 66, 317-322.	2.3	5
112	DISCORDANCE FOR RETINITIS PIGMENTOSA IN TWO MONOZYGOTIC TWIN PAIRS. Retina, 2011, 31, 1164-1169.	1.7	5
113	Identification of two de novo mutations responsible for type I antithrombin deficiency. Thrombosis and Haemostasis, 2012, 107, 187-189.	3.4	4
114	The deletion F508 is the major gene mutation in a representative Belgian cystic fibrosis population. Human Genetics, 1990, 85, 395-396.	3.8	3
115	Nonsense mutation Arg197stop in a Dutch family with type 1 hereditary antithrombin (AT) deficiency causing thrombophilia. Thrombosis Research, 1995, 78, 251-254.	1.7	3
116	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.	1.5	3
117	Clinical variability in neurohepatic syndrome due to combined mitochondrial DNA depletion and Gaucher disease. Molecular Genetics and Metabolism Reports, 2014, 1, 223-231.	1.1	2
118	Convert your favorite protein modeling program into a mutation predictor: â€œMODICTâ€• BMC Bioinformatics, 2016, 17, 425.	2.6	2
119	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.	0.6	1
120	Identification of two novel mutations in the cystic fibrosis gene: 1898 + 3Aâ†'C and 2711delT. Human Mutation, 1995, 6, 188-189.	2.5	0
121	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.	7.4	0
122	Genes and infertility. , 0, , 113-126.		0
123	Severe male factor. , 2008, , 343-356.		0
124	Severe male factor: Genetic consequences and recommendations for genetic testing. , 2012, , 324-335.		0