Fortunato Lonardo

List of Publications by Year
in descending order

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50	915	516710	477307
papers	citations	16	29
		h-index	g-index

53	53	53	1446
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Cryptic deletions are a common finding in "balanced" reciprocal and complex chromosome rearrangements: a study of 59 patients. Journal of Medical Genetics, 2007, 44, 750-762.	3.2	244
2	Expanding the clinical phenotype of individuals with a 3-bp in-frame deletion of the NF1 gene (c.2970_2972del): an update of genotypeâ€“phenotype correlation. Genetics in Medicine, 2019, 21, 867-876.	2.4	62
3	Paternal mosaicism for a COL1A1 dominant mutation (Î±1 Ser-415) causes recurrent osteogenesis imperfecta. Human Mutation, 1993, 2, 196-204.	2.5	40
4	Genotypeâ€“phenotype correlations in recessive titinopathies. Genetics in Medicine, 2020, 22, 2029-2040.	2.4	35
5	Contiguous gene syndrome due to an interstitial deletion in Xp22.3 in a boy with ichthyosis, chondrodysplasia punctata, mental retardation and ADHD. European Journal of Medical Genetics, 2007, 50, 301-308.	1.3	34
6	Majewski osteodysplastic primordial dwarfism type II (MOPD II) syndrome previously diagnosed as Seckel syndrome: Report of a novel mutation of the PCNT gene. American Journal of Medical Genetics, Part A, 2009, 149A, 2452-2456.	1.2	33
7	SNORD116 deletions cause Praderâ€“Willi syndrome with a mild phenotype and macrocephaly. Clinical Genetics, 2017, 92, 440-443.	2.0	31
8	Prenatal diagnosis of femurâ€“fibulaâ€“ulna complex by ultrasonography in a male fetus at 24 weeks of gestation. Prenatal Diagnosis, 1994, 14, 502-505.	2.3	30
9	Prenatal ultrasound diagnosis of a case of Pfeiffer syndrome without cloverleaf skull and review of the literature. Prenatal Diagnosis, 2004, 24, 918-922.	2.3	28
10	Prenatal ultrasound diagnosis of macroglossia in the Wiedemann-Beckwith syndrome. Prenatal Diagnosis, 1988, 8, 79-81.	2.3	25
11	Identification of 14 rare marker chromosomes and derivatives by spectral karyotyping in prenatal and postnatal diagnosis. American Journal of Medical Genetics, Part A, 2004, 127A, 144-148.	1.2	23
12	Gastroschisis in two sibs with abdominal hernia in maternal grandfather and greatgrandfather. American Journal of Medical Genetics Part A, 1985, 21, 405-407.	2.4	22
13	Narrowing the candidate region of Albright hereditary osteodystrophy-like syndrome by deletion mapping in a patient with an unbalanced cryptic translocation t(2;6)(q37.3;q26). American Journal of Medical Genetics Part A, 2003, 122A, 261-265.	2.4	20
14	Prenatal diagnosis of Mohr syndrome by ultrasonography. Prenatal Diagnosis, 1985, 5, 415-418.	2.3	19
15	Sayâ€“Barberâ€“Biesekerâ€“Youngâ€“Simpson syndrome and Genitopatellar syndrome: Lumping or splitting?. Clinical Genetics, 2019, 95, 253-261.	2.0	19
16	Prenatal ultrasound diagnosis of cloacal exstrophy associated with myelocystocele complex by the â€˜elephant trunk-likeâ€™ image and review of the literature. Prenatal Diagnosis, 2005, 25, 394-397.	2.3	18
17	Mucopolysaccharidosis type II in a female patient with a reciprocal X;9 translocation and skewed X chromosome inactivation. American Journal of Medical Genetics, Part A, 2014, 164, 2627-2632.	1.2	17
18	First evidence of Smithâ€“Magenis syndrome in mother and daughter due to a novel RAI mutation. American Journal of Medical Genetics, Part A, 2017, 173, 231-238.	1.2	17

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19	Small 4p16.3 deletions: Three additional patients and review of the literature. American Journal of Medical Genetics, Part A, 2018, 176, 2501-2508.	1.2	16
20	A spectrum of molecular variation in a cohort of Italian families with trimethylaminuria: Identification of three novel mutations of the FM03 gene. Molecular Genetics and Metabolism, 2006, 88, 192-195.	1.1	15
21	A family with X-linked recessive fusion of metacarpals IV and V. American Journal of Medical Genetics Part A, 2004, 124A, 407-410.	2.4	14
22	A case of autism with an interstitial 1q deletion (1q23.3â€24.2) and a de novo translocation of chromosomes 1q and 5q. American Journal of Medical Genetics, Part A, 2007, 143A, 2733-2737.	1.2	13
23	Centrosome Linkerâ€“induced Tetraploid Segregation Errors Link Rhabdoid Phenotypes and Lethal Colorectal Cancers. Molecular Cancer Research, 2018, 16, 1385-1395.	3.4	13
24	Clinical heterogeneity of Kabuki syndrome in a cohort of Italian patients and review of the literature. European Journal of Pediatrics, 2022, 181, 171-187.	2.7	13
25	Diagnostic application of a capture based <scp>NGS</scp> test for the concurrent detection of variants in sequence and copy number as well as <scp>LOH</scp>. Clinical Genetics, 2018, 93, 545-556.	2.0	12
26	Diagnostic issues faced by a rare disease healthcare network during Covid-19 outbreak: data from the Campania Rare Disease Registry. Journal of Public Health, 2022, 44, 586-594.	1.8	12
27	Clinical, cytogenetic and molecular-cytogenetic characterization of a patient with a de novo tandem proximal-intermediate duplication of 16q and review of the literature. , 2011, 155, 769-777.		11
28	Baseline Inhibin B Levels for Diagnosis of Central Precocious Puberty in Girls. Hormone Research in Paediatrics, 2013, 80, 207-212.	1.8	8
29	Novel findings in a patient with Weaver or a Weaver-like syndrome. , 1996, 63, 378-381.		7
30	Prenatal diagnosis of 46,XX testicular DSD. Molecular, cytogenetic, molecularâ€cytogenetic, and ultrasonographic evaluation. Prenatal Diagnosis, 2009, 29, 998-1001.	2.3	7
31	A novel OTOA mutation in an Italian family with hearing loss. Gene Reports, 2017, 9, 111-114.	0.8	7
32	"A Case of Autosomal Recessive Form of Cranio-Metaphyseal Dysplasia with Unusual Features and with Bone Fragility". Journal of Medical Imaging and Radiation Oncology, 1987, 31, 79-82.	0.6	6
33	Al-Awadi/Raas-Rothschild syndrome: Two new cases and review. American Journal of Medical Genetics, Part A, 2007, 143A, 3169-3174.	1.2	6
34	Delineation of MidXq28â€duplication syndrome distal to MECP2 and proximal to RAB39B genes. Clinical Genetics, 2019, 96, 246-253.	2.0	6
35	Clinical and molecular spectrum of Wiedemann-Steiner syndrome, an emerging member of the chromatinopathy family. World Journal of Medical Genetics, 2020, 9, 1-11.	1.0	5
36	Science, art, and mistery in the statues and in the anatomical machines of the prince of sansevero: The masterpieces of the â€œSansevero Chapelâ€. American Journal of Medical Genetics, Part A, 2013, 161, 2920-2929.	1.2	4

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37	The Salernitan school of medicine: Women, men, and children. A syndromological review of the oldest medical school in the western world. American Journal of Medical Genetics, Part A, 2013, 161, 809-816.	1.2	4
38	A ZFHX4 mutation associated with a recognizable neuropsychological and facial phenotype. European Journal of Medical Genetics, 2021, 64, 104321.	1.3	4
39	Clinical and molecular characterizations of 11 new patients with type 1 Feingold syndrome: Proposal for selecting diagnostic criteria and further genetic testing in patients with severe phenotype. American Journal of Medical Genetics, Part A, 2021, 185, 1204-1210.	1.2	4
40	Mental retardation, Robin sequence, and brachydactyly: Further confirmation of a new syndrome. American Journal of Medical Genetics Part A, 2004, 126A, 204-207.	2.4	2
41	Maternal uniparental disomy of the chromosome 14: need for growth hormone provocative tests also when a deficiency is not suspected. BMJ Case Reports, 2019, 12, e228662.	0.5	2
42	Report of Three Cases with Tricho-Rhino-Phalangeal Syndrome Type I (Two Cases) and Type II (One Case). Journal of Medical Imaging and Radiation Oncology, 1988, 32, 338-342.	0.6	1
43	The promise of non-invasive prenatal testing needs to be monitored scientifically. BMJ, The, 2015, 350, h2518-h2518.	6.0	1
44	Prenatal diagnosis of a novel pathogenic variation in the ACAN gene presenting with isolated shortening of fetal long bones in the second trimester of gestation: a case report. BMC Pregnancy and Childbirth, 2021, 21, 459.	2.4	1
45	De Novo Inverted Duplication Deletion of 4p in a 14-Week-Old Male Fetus Aborted Due to Multiple Anomalies. Journal of Pediatric Genetics, 2021, 10, 245-249.	0.7	1
46	A case of short-rib syndrome without polydactyly in a stillborn: a new type?. Birth Defects: Original Article Series, 1996, 30, 95-101.	0.1	1
47	Clinical Characterization of a 6-Year-Old Patient with Autism and Two Adjacent Duplications on 10q11.22q11.23. A Case Report. Children, 2021, 8, 518.	1.5	0
48	Genomic microarrays in prenatal diagnosis. World Journal of Medical Genetics, 2013, 3, 14.	1.0	0
49	Two sibs affected by Pendred's syndrome in a family with recurrent goiter. Minerva Endocrinologica, 1987, 12, 29-32.	1.8	0
50	The Italian National External Quality Assessment Program in Cytogenetics: 4 years of activity (2013-2016) following the introduction of poor performance criteria. Annali Dell'Istituto Superiore Di Sanita, 2018, 54, 109-116.	0.4	0