Claudia Izzi

List of Publications by Year in descending order

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51 papers	3,687 citations	257450 24 h-index	182427 51 g-index
55 all docs	55 docs citations	55 times ranked	5555 citing authors

#	Article	IF	Citations
1	Lessons From the Clinic: ADPKD Genetic Test Unraveling Severe Phenotype, Intrafamilial Variability, and New, Rare Causing Genotype. Kidney International Reports, 2022, 7, 895-898.	0.8	3
2	Copy Number Variant Analysis and Genome-wide Association Study Identify Loci with Large Effect for Vesicoureteral Reflux. Journal of the American Society of Nephrology: JASN, 2021, 32, 805-820.	6.1	17
3	Expanding the variability of the ADPKD-GANAB clinical phenotype inÂa family of Italian ancestry. Journal of Nephrology, 2021, , 1.	2.0	5
4	Outcomes in pregnancies with a confined placental mosaicism and implications for prenatal screening using cell-free DNA. Genetics in Medicine, 2020, 22, 309-316.	2.4	73
5	Variable Expressivity of HNF1B Nephropathy, From Renal Cysts and Diabetes to Medullary Sponge Kidney Through Tubulo-interstitial Kidney Disease. Kidney International Reports, 2020, 5, 2341-2350.	0.8	21
6	An international cohort study of autosomal dominant tubulointerstitial kidney disease due to mutations identifies distinct clinical subtypes. Kidney International, 2020, 98, 1589-1604.	5.2	27
7	Genetic and Clinical Predictors of Age of ESKD in Individuals With Autosomal Dominant Tubulointerstitial Kidney Disease Due to UMOD Mutations. Kidney International Reports, 2020, 5, 1472-1485.	0.8	30
8	Testicular Involvement is a Hallmark of Apo A-I Leu75Pro Mutation Amyloidosis. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e4758-e4766.	3.6	4
9	Outcomes in Pregnancies With a Confined Placental Mosaicism and Implications for Prenatal Screening Using Cell-Free DNA. Obstetrical and Gynecological Survey, 2020, 75, 397-398.	0.4	1
10	The genetic architecture of membranous nephropathy and its potential to improve non-invasive diagnosis. Nature Communications, 2020, 11, 1600.	12.8	120
11	Autosomal Dominant Tubulointerstitial Kidney Disease with Adult Onset due to a Novel Renin Mutation Mapping in the Mature Protein. Scientific Reports, 2019, 9, 11601.	3.3	19
12	Prenatal findings in oralâ€facialâ€digital syndrome type VI: Report of three cases and literature review. Prenatal Diagnosis, 2019, 39, 652-655.	2.3	1
13	Discrepant molecular and clinical diagnoses in Beckwith-Wiedemann and Silver-Russell syndromes. Genetical Research, 2019, 101, e3.	0.9	17
14	The copy number variation landscape of congenital anomalies of the kidney and urinary tract. Nature Genetics, 2019, 51, 117-127.	21.4	144
15	Identification of novel mutations in patients with fibrinogen disorders and genotype/phenotype correlations. Blood Transfusion, 2019, 17, 247-254.	0.4	3
16	A new case report of severe mucopolysaccharidosis type VII: diagnosis, treatment with haematopoietic cell transplantation and prenatal diagnosis in a second pregnancy. Italian Journal of Pediatrics, 2018, 44, 128.	2.6	12
17	Expanding the phenotype of MED 17 mutations: Description of two new cases and review of the literature. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 687-690.	1.7	7
18	Genetic Drivers of Kidney Defects in the DiGeorge Syndrome. New England Journal of Medicine, 2017, 376, 742-754.	27.0	120

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19	Exome-wide Association Study Identifies GREB1L Mutations in Congenital Kidney Malformations. American Journal of Human Genetics, 2017, 101, 789-802.	6.2	63
20	Comparative X-ray morphometry of prenatal osteogenesis imperfecta type 2 and thanatophoric dysplasia: a contribution to prenatal differential diagnosis. Radiologia Medica, 2017, 122, 880-891.	7.7	3
21	Congenital Anomalies of the Kidney and the Urinary Tract (CAKUT). Giornale De Techniche Nefrologiche & Dialitiche, 2016, 28, 79-82.	0.1	0
22	Deciphering Variability of PKD1 and PKD2 in an Italian Cohort of 643 Patients with Autosomal Dominant Polycystic Kidney Disease (ADPKD). Scientific Reports, 2016, 6, 30850.	3.3	28
23	Interpreting mosaicism in chorionic villi: results of a monocentric series of 1001 mosaics in chorionic villi with follow-up amniocentesis. Prenatal Diagnosis, 2015, 35, 1117-1127.	2.3	134
24	Tubulointerstitial nephritis is a dominant feature of hereditary apolipoprotein A-I amyloidosis. Kidney International, 2015, 87, 1223-1229.	5.2	28
25	Uromodulin: from monogenic to multifactorial diseases: FIGUREÂ1:. Nephrology Dialysis Transplantation, 2015, 30, 1250-1256.	0.7	57
26	The Case | Cystic renal disease, nephrogenic diabetes insipidus, and polycytemia. Kidney International, 2014, 86, 863-864.	5.2	2
27	Fetal abdominal wall defects. Best Practice and Research in Clinical Obstetrics and Gynaecology, 2014, 28, 391-402.	2.8	77
28	Discovery of new risk loci for IgA nephropathy implicates genes involved in immunity against intestinal pathogens. Nature Genetics, 2014, 46, 1187-1196.	21.4	505
29	Thanatophoric dysplasia. Correlation among bone X-ray morphometry, histopathology, and gene analysis. Skeletal Radiology, 2014, 43, 1205-1215.	2.0	7
30	Complex rearrangement of the exon 6 genomic region among Opitz G/BBB Syndrome MID1 alterations. European Journal of Medical Genetics, 2013, 56, 404-410.	1.3	9
31	Mutation Spectrum inRAB3GAP1,RAB3GAP2, andRAB18and Genotype-Phenotype Correlations in Warburg Micro Syndrome and Martsolf Syndrome. Human Mutation, 2013, 34, 686-696.	2.5	114
32	Prenatal presentation and postnatal evolution of a patient with Jansen metaphyseal dysplasia with a novel missense mutation in PTH1R. American Journal of Medical Genetics, Part A, 2013, 161, 2614-2619.	1.2	11
33	Mutations in DSTYK and Dominant Urinary Tract Malformations. New England Journal of Medicine, 2013, 369, 621-629.	27.0	119
34	Geographic Differences in Genetic Susceptibility to IgA Nephropathy: GWAS Replication Study and Geospatial Risk Analysis. PLoS Genetics, 2012, 8, e1002765.	3.5	301
35	Copy-Number Disorders Are a Common Cause of Congenital Kidney Malformations. American Journal of Human Genetics, 2012, 91, 987-997.	6.2	201
36	Urinary secretion and extracellular aggregation of mutant uromodulin isoforms. Kidney International, 2012, 81, 769-778.	5.2	20

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37	Genome-wide association study identifies susceptibility loci for IgA nephropathy. Nature Genetics, 2011, 43, 321-327.	21.4	528
38	Alport syndrome and leiomyomatosis: the first deletion extending beyond COL4A6 intron 2. Pediatric Nephrology, 2011, 26, 717-724.	1.7	27
39	The Case â^£ Familial occurrence of retinitis pigmentosa, deafness, and nephropathy. Kidney International, 2011, 79, 691-692.	5.2	10
40	Type I hyperprolinemia: genotype/phenotype correlations. Human Mutation, 2010, 31, 961-965.	2.5	26
41	Identification and characterization of seven novel mutations of elastin gene in a cohort of patients affected by supravalvular aortic stenosis. European Journal of Human Genetics, 2010, 18, 317-323.	2.8	51
42	IgA nephropathy--the case for a genetic basis becomes stronger. Nephrology Dialysis Transplantation, 2010, 25, 336-338.	0.7	11
43	A Recessive Gene for Primary Vesicoureteral Reflux Maps to Chromosome 12p11-q13. Journal of the American Society of Nephrology: JASN, 2009, 20, 1633-1640.	6.1	42
44	Renal outcome in patients with congenital anomalies of the kidney and urinary tract. Kidney International, 2009, 76, 528-533.	5.2	309
45	Localization of a Gene for Nonsyndromic Renal Hypodysplasia to Chromosome 1p32-33. American Journal of Human Genetics, 2007, 80, 539-549.	6.2	33
46	IgA Nephropathy: The Presence of Familial Disease Does Not Confer an Increased Risk for Progression. American Journal of Kidney Diseases, 2006, 47, 761-769.	1.9	23
47	Renal Apolipoprotein A-I Amyloidosis: A Rare and Usually Ignored Cause of Hereditary Tubulointerstitial Nephritis. Journal of the American Society of Nephrology: JASN, 2005, 16, 3680-3686.	6.1	83
48	Familial Vesicoureteral Reflux: Testing Replication of Linkage in Seven New Multigenerational Kindreds. Journal of the American Society of Nephrology: JASN, 2005, 16, 1781-1787.	6.1	56
49	Uromodulin storage diseases: Clinical aspects and mechanisms. American Journal of Kidney Diseases, 2004, 44, 987-999.	1.9	123
50	Candidate gene analysis of the human metabotropic glutamate receptor type 4 (GRM4) in patients with juvenile myoclonic epilepsy. , 2003, 123B, 59-63.		14
51	Sequencing of theGRIK1 gene in patients with juvenile absence epilepsy does not reveal mutations affecting receptor structure. American Journal of Medical Genetics Part A, 2002, 114, 354-359.	2.4	21