Lorenzo D Botto

List of Publications by Year
in descending order

100	8,104	117625	48315
papers	citations	34 h-index	88 g-index

104	104	104	8341
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Patterns of multiple congenital anomalies in the National Birth Defect Prevention Study: Challenges and insights. Birth Defects Research, 2023, 115, 43-55.	1.5	4
2	Maternal Smoking and Congenital Heart Defects, National Birth Defects Prevention Study, 1997-2011. Journal of Pediatrics, 2022, 240, 79-86.e1.	1.8	12
3	Survival of infants born with esophageal atresia among 24 international birth defects surveillance programs. Birth Defects Research, 2021, 113, 945-957.	1.5	8
4	Paternal genetic variants and risk of obstructive heart defects: A parent-of-origin approach. PLoS Genetics, 2021, 17, e1009413.	3.5	2
5	Modification of the association between diabetes and birth defects by obesity, National Birth Defects Prevention Study, 1997â€“2011. Birth Defects Research, 2021, 113, 1084-1097.	1.5	9
6	Skeletal dysplasias in art and antiquities: A cultural journey through genes, environment, and chance. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 199-212.	1.6	1
7	Global birth defects app: An innovative tool for describing and coding congenital anomalies at birth in low resource settings. Birth Defects Research, 2021, 113, 1057-1073.	1.5	6
8	Chondrodysplasia and growth failure in children after early hematopoietic stem cell transplantation for nonâ€oncologic disorders. American Journal of Medical Genetics, Part A, 2021, 185, 517-527.	1.2	3
9	Specific birth defects in pregnancies of women with diabetes: National Birth Defects Prevention Study, 1997â€“2011. American Journal of Obstetrics and Gynecology, 2020, 222, 176.e1-176.e11.	1.3	84
10	Clinical and biochemical outcomes of patients with medium-chain acyl-CoA dehydrogenase deficiency. Molecular Genetics and Metabolism, 2020, 129, 13-19.	1.1	21
11	Comparative Serum Analyses Identify Cytokines and Hormones Commonly Dysregulated as Well as Implicated in Promoting Osteolysis in MMP-2-Deficient Mice and Children. Frontiers in Physiology, 2020, 11, 568718.	2.8	1
12	From cause to care: Can a triple approach to better population data improve the global outlook of congenital heart disease?. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 23-35.	1.6	4
13	A genome-wide association study implicates the BMP7 locus as a risk factor for nonsyndromic metopic craniosynostosis. Human Genetics, 2020, 139, 1077-1090.	3.8	24
14	Associations between PM2.5 and risk of preterm birth among liveborn infants. Annals of Epidemiology, 2019, 39, 46-53.e2.	1.9	15
15	Prenatal diagnosis and prevalence of critical congenital heart defects: an international retrospective cohort study. BMJ Open, 2019, 9, e028139.	1.9	126
16	Maternal exposure to outdoor air pollution and congenital limb deficiencies in the National Birth Defects Prevention Study. Environmental Research, 2019, 179, 108716.	7.5	14
17	Projected Changes in Maternal Heat Exposure During Early Pregnancy and the Associated Congenital Heart Defect Burden in the United States. Journal of the American Heart Association, 2019, 8, e010995.	3.7	41
18	Risk of gastroschisis with maternal genitourinary infections: the US National birth defects prevention study 1997â€“2011. BMJ Open, 2019, 9, e026297.	1.9	16

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19	Targeted gene panel sequencing for the rapid diagnosis of acutely ill infants. Molecular Genetics & Genomic Medicine, 2019, 7, e00796.	1.2	34
20	Join World Birth Defects Day. Pediatric Research, 2019, 86, 3-4.	2.3	12
21	Identification of fibrinogen as a natural inhibitor of MMP-2. Scientific Reports, 2019, 9, 4340.	3.3	15
22	Risk factors associated with the development of doubleâ€inlet ventricle congenital heart disease. Birth Defects Research, 2019, 111, 640-648.	1.5	10
23	An evolutionary and developmental biology approach to gastroschisis. Birth Defects Research, 2019, 111, 294-311.	1.5	34
24	SLC35A2â€CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. Human Mutation, 2019, 40, 908-925.	2.5	39
25	Expanding the genetic and clinical spectrum of the NONOâ€associated Xâ€linked intellectual disability syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 792-796.	1.2	21
26	Application of quality indicators to data from the National Network of Congenital Anomalies of Argentina. Birth Defects Research, 2019, 111, 333-340.	1.5	2
27	A proposal for the systematic assessment of data quality indicators in birth defects surveillance. Birth Defects Research, 2019, 111, 324-332.	1.5	2
28	Congenital anomalies and associated risk factors in a Saudi population: a cohort study from pregnancy to age 2 years. BMJ Open, 2019, 9, e026351.	1.9	17
29	Brief Report: Pediatrician Perspectives Regarding Genetic Evaluations of Children with Autism Spectrum Disorder. Journal of Autism and Developmental Disorders, 2019, 49, 794-808.	2.7	13
30	Osteoporosis and skeletal dysplasia caused by pathogenic variants in SGMS2. JCI Insight, 2019, 4, .	5.0	47
31	Triple surveillance: a proposal for an integrated strategy to support and accelerate birth defect prevention. Annals of the New York Academy of Sciences, 2018, 1414, 126-136.	3.8	19
32	Does Maternal Exposure to Secondhand Tobacco Smoke During Pregnancy Increase the Risk for Preterm or Small-for-Gestational Age Birth?. Maternal and Child Health Journal, 2018, 22, 1418-1429.	1.5	17
33	Intracranial Calcifications in Young Children. Seminars in Pediatric Neurology, 2018, 26, 135-139.	2.0	3
34	Value of sharing and networking among birth defects surveillance programs: an ICBDSR perspective. Journal of Community Genetics, 2018, 9, 411-415.	1.2	8
35	Early Childhood Inpatient Costs of Critical Congenital Heart Disease. Journal of Pediatrics, 2018, 203, 371-379.e7.	1.8	22
36	Delineation of the 9q31 deletion syndrome: Genomic microarray characterization of two patients with overlapping deletions. American Journal of Medical Genetics, Part A, 2018, 176, 2901-2906.	1.2	4

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37	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. American Journal of Human Genetics, 2018, 103, 666-678.	6.2	87
38	Case Definitions for Conditions Identified by Newborn Screening Public Health Surveillance. International Journal of Neonatal Screening, 2018, 4, 16.	3.2	17
39	From cause to care: Triple surveillance for better outcomes in birth defects and rare diseases. European Journal of Medical Genetics, 2018, 61, 551-555.	1.3	8
40	Development and Utility of a Birth Defects Surveillance Toolkit. Journal of Global Health Perspectives, 2018, 0, .	0.3	0
41	Mild orotic aciduria in UMPS heterozygotes: a metabolic finding without clinical consequences. Journal of Inherited Metabolic Disease, 2017, 40, 423-431.	3.6	14
42	Costs, mortality, and hospital usage in relation to prenatal diagnosis in dâ€transposition of the great arteries. Birth Defects Research, 2017, 109, 262-270.	1.5	5
43	Epidemiology and Prognosis of Congenital Diaphragmatic Hernia: A Populationâ€Based Cohort Study in Utah. Birth Defects Research, 2017, 109, 1451-1459.	1.5	66
44	Maternal Exposure to Nitrogen Dioxide, Intake of Methyl Nutrients, and Congenital Heart Defects in Offspring. American Journal of Epidemiology, 2017, 186, 719-729.	3.4	24
45	Etiology and clinical presentation of birth defects: population based study. BMJ: British Medical Journal, 2017, 357, j2249.	2.3	125
46	Bayesian multinomial probit modeling of daily windows of susceptibility for maternal PM_{2.5} exposure and congenital heart defects. Statistics in Medicine, 2016, 35, 2786-2801.	1.6	19
47	Clinical presentation and survival in a populationâ€based cohort of infants with gastroschisis in Utah, 1997â€“2011. American Journal of Medical Genetics, Part A, 2016, 170, 306-315.	1.2	19
48	Diagnosis, Treatment, and Clinical Outcome of Patients with Mitochondrial Trifunctional Protein/Long-Chain 3-Hydroxy Acyl-CoA Dehydrogenase Deficiency. JIMD Reports, 2016, 31, 63-71.	1.5	25
49	Associations between maternal periconceptional exposure to secondhand tobacco smoke and major birth defects. American Journal of Obstetrics and Gynecology, 2016, 215, 613.e1-613.e11.	1.3	51
50	Loss-of-Function Mutations in ELMO2 Cause Intraosseous Vascular Malformation by Impeding RAC1 Signaling. American Journal of Human Genetics, 2016, 99, 299-317.	6.2	23
51	Biallelic Variants in UBA5 Reveal that Disruption of the UFM1 Cascade Can Result in Early-Onset Encephalopathy. American Journal of Human Genetics, 2016, 99, 695-703.	6.2	87
52	Databases for Congenital Heart Defect Public Health Studies Across the Lifespan. Journal of the American Heart Association, 2016, 5, .	3.7	24
53	Effect of dietary lysine restriction and arginine supplementation in two patients with pyridoxine-dependent epilepsy. Molecular Genetics and Metabolism, 2016, 118, 167-172.	1.1	32
54	Elevated body mass index and decreased diet quality among women and risk of birth defects in their offspring. Birth Defects Research Part A: Clinical and Molecular Teratology, 2016, 106, 164-171.	1.6	14

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55	Lower rate of selected congenital heart defects with better maternal diet quality: a population-based study. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2016, 101, 43-49.	2.8	32
56	Evaluating cost and resource use associated with pulse oximetry screening for critical congenital heart disease: Empiric estimates and sources of variation. Birth Defects Research Part A: Clinical and Molecular Teratology, 2015, 103, 962-971.	1.6	9
57	Maternal periconceptional alcohol consumption and congenital heart defects. Birth Defects Research Part A: Clinical and Molecular Teratology, 2015, 103, 617-629.	1.6	27
58	Risk factors for Dandyâ€“Walker malformation: A populationâ€based assessment. American Journal of Medical Genetics, Part A, 2015, 167, 2009-2016.	1.2	14
59	Turner Syndrome in Girls Presenting with Coarctation of the Aorta. Journal of Pediatrics, 2015, 167, 1062-1066.	1.8	33
60	Reflections on the etiology of structural birth defects: Established teratogens and risk factors. Birth Defects Research Part A: Clinical and Molecular Teratology, 2015, 103, 652-655.	1.6	13
61	Building capacity for birth defects surveillance in Africa: Implementation of an intermediate birth defects surveillance workshop. Journal of Global Health Perspectives, 2015, 2015, .	0.3	11
62	Better Diet Quality before Pregnancy Is Associated with Reduced Risk of Gastroschisis in Hispanic Women. Journal of Nutrition, 2014, 144, 1781-1786.	2.9	17
63	Developing a public health-tracking system for follow-up of newborn screening metabolic conditions: a four-state pilot project structure and initial findings. Genetics in Medicine, 2014, 16, 484-490.	2.4	22
64	Maternal Exposure to Criteria Air Pollutants and Congenital Heart Defects in Offspring: Results from the National Birth Defects Prevention Study. Environmental Health Perspectives, 2014, 122, 863-872.	6.0	82
65	Maternal Butalbital Use and Selected Defects in the National Birth Defects Prevention Study. Headache, 2014, 54, 54-66.	3.9	23
66	Maternal intake of vitamin E and birth defects, national birth defects prevention study, 1997 to 2005. Birth Defects Research Part A: Clinical and Molecular Teratology, 2014, 100, 647-657.	1.6	14
67	Laterality defects in the national birth defects prevention study (1998â€“2007): Birth prevalence and descriptive epidemiology. American Journal of Medical Genetics, Part A, 2014, 164, 2581-2591.	1.2	145
68	Congenital heart defects after maternal fever. American Journal of Obstetrics and Gynecology, 2014, 210, 359.e1-359.e11.	1.3	29
69	Cancer Risk in Children and Adolescents with Birth Defects: A Population-Based Cohort Study. PLoS ONE, 2013, 8, e69077.	2.5	67
70	Selected birth defects data from populationâ€based birth defects surveillance programs in the United States, 2005â€“2009: Featuring critical congenital heart defects targeted for pulse oximetry screening. Birth Defects Research Part A: Clinical and Molecular Teratology, 2012, 94, 970-983.	1.6	43
71	Newborn screening for critical congenital heart disease: Essential public health roles for birth defects monitoring programs. Birth Defects Research Part A: Clinical and Molecular Teratology, 2012, 94, 965-969.	1.6	16
72	Analysis of selected maternal exposures and nonâ€syndromic atrioventricular septal defects in the National Birth Defects Prevention Study, 1997â€“2005.. American Journal of Medical Genetics, Part A, 2012, 158A, 2447-2455.	1.2	44

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73	Lack of periconceptional vitamins or supplements that contain folic acid and diabetes mellitusâ€“associated birth defects. American Journal of Obstetrics and Gynecology, 2012, 206, 218.e1-218.e13.	1.3	78
74	The Contribution of Chromosomal Abnormalities to Congenital Heart Defects: A Population-Based Study. Pediatric Cardiology, 2011, 32, 1147-1157.	1.3	161
75	Acardia: Epidemiologic findings and literature review from the International Clearinghouse for Birth Defects Surveillance and Research. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2011, 157, 262-273.	1.6	6
76	Is gastroschisis truly a sporadic defect? Familial cases of gastroschisis in Utah, 1997 to 2008. Birth Defects Research Part A: Clinical and Molecular Teratology, 2011, 91, 873-878.	1.6	24
77	Acetaminophen Use in Pregnancy and Risk of Birth Defects. Obstetrics and Gynecology, 2010, 115, 109-115.	2.4	94
78	Surveillance of adverse fetal effects of medications (SAFE-Med): Findings from the International Clearinghouse of Birth Defects Surveillance and Research. Reproductive Toxicology, 2010, 29, 433-442.	2.9	33
79	Association between prepregnancy body mass index and congenital heart defects. American Journal of Obstetrics and Gynecology, 2010, 202, 51.e1-51.e10.	1.3	106
80	Maternal use of bupropion and risk for congenital heart defects. American Journal of Obstetrics and Gynecology, 2010, 203, 52.e1-52.e6.	1.3	78
81	How valid are the rates of Down syndrome internationally? Findings from the International Clearinghouse for Birth Defects Surveillance and Research. American Journal of Medical Genetics, Part A, 2010, 152A, 1670-1680.	1.2	34
82	Developing a research and public health agenda for gastroschisis: How do we bridge the gap between what is known and what is not?. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2008, 148C, 155-161.	1.6	20
83	Diabetes mellitus and birth defects. American Journal of Obstetrics and Gynecology, 2008, 199, 237.e1-237.e9.	1.3	530
84	Maternal Smoking and Congenital Heart Defects. Pediatrics, 2008, 121, e810-e816.	2.1	202
85	Noninherited Risk Factors and Congenital Cardiovascular Defects: Current Knowledge. Circulation, 2007, 115, 2995-3014.	1.6	663
86	Flavors in Geneâ€“Environment Interactions. Epidemiology, 2007, 18, 431-432.	2.7	6
87	Seeking causes: Classifying and evaluating congenital heart defects in etiologic studies. Birth Defects Research Part A: Clinical and Molecular Teratology, 2007, 79, 714-727.	1.6	367
88	Maternal plasma and erythrocyte folate levels and risk of oral clefts in Utah. FASEB Journal, 2007, 21, .	0.5	0
89	Fostering International Collaboration in Birth Defects Research and Prevention: A Perspective From the International Clearinghouse for Birth Defects Surveillance and Research. American Journal of Public Health, 2006, 96, 774-780.	2.7	34
90	Trends of selected malformations in relation to folic acid recommendations and fortification: An international assessment. Birth Defects Research Part A: Clinical and Molecular Teratology, 2006, 76, 693-705.	1.6	87

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91	International retrospective cohort study of neural tube defects in relation to folic acid recommendations: are the recommendations working?. BMJ: British Medical Journal, 2005, 330, 571.	2.3	205
92	Vitamin supplements and the risk for congenital anomalies other than neural tube defects. American Journal of Medical Genetics Part A, 2004, 125C, 12-21.	2.4	192
93	Do multivitamin or folic acid supplements reduce the risk for congenital heart defects? Evidence and gaps. American Journal of Medical Genetics Part A, 2003, 121A, 95-101.	2.4	110
94	Decreasing the burden of congenital heart anomalies: an epidemiologic evaluation of risk factors and survival. Progress in Pediatric Cardiology, 2003, 18, 111-121.	0.4	117
95	A Population-Based Study of the 22q11.2 Deletion: Phenotype, Incidence, and Contribution to Major Birth Defects in the Population. Pediatrics, 2003, 112, 101-107.	2.1	606
96	Maternal Fever, Multivitamin Use, and Selected Birth Defects: Evidence of Interaction?. Epidemiology, 2002, 13, 485-488.	2.7	91
97	Congenital Heart Defects, Maternal Febrile Illness, and Multivitamin Use: A Population-Based Study. Epidemiology, 2001, 12, 485-490.	2.7	112
98	Mortality Associated With Congenital Heart Defects in the United States. Circulation, 2001, 103, 2376-2381.	1.6	469
99	5, 10-Methylenetetrahydrofolate Reductase Gene Variants and Congenital Anomalies: A HuGE Review. American Journal of Epidemiology, 2000, 151, 862-877.	3.4	871
100	Neural-Tube Defects. New England Journal of Medicine, 1999, 341, 1509-1519.	27.0	800