

Lorenzo D Botto

List of Publications by Year in descending order

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Version: 2024-02-01

100
papers

8,104
citations

117625

34
h-index

48315

88
g-index

104
all docs

104
docs citations

104
times ranked

8341
citing authors

#	ARTICLE	IF	CITATIONS
1	5, 10-Methylenetetrahydrofolate Reductase Gene Variants and Congenital Anomalies: A HuGE Review. American Journal of Epidemiology, 2000, 151, 862-877.	3.4	871
2	Neural-Tube Defects. New England Journal of Medicine, 1999, 341, 1509-1519.	27.0	800
3	Noninherited Risk Factors and Congenital Cardiovascular Defects: Current Knowledge. Circulation, 2007, 115, 2995-3014.	1.6	663
4	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
5	Diabetes mellitus and birth defects. American Journal of Obstetrics and Gynecology, 2008, 199, 237.e1-237.e9.	1.3	530
6	Mortality Associated With Congenital Heart Defects in the United States. Circulation, 2001, 103, 2376-2381.	1.6	469
7	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
8	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
9	Maternal Smoking and Congenital Heart Defects. Pediatrics, 2008, 121, e810-e816.	2.1	202
10	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
11	The Contribution of Chromosomal Abnormalities to Congenital Heart Defects: A Population-Based Study. Pediatric Cardiology, 2011, 32, 1147-1157.	1.3	161
12	Laterality defects in the national birth defects prevention study (1998-2007): Birth prevalence and descriptive epidemiology. American Journal of Medical Genetics, Part A, 2014, 164, 2581-2591.	1.2	145
13	Prenatal diagnosis and prevalence of critical congenital heart defects: an international retrospective cohort study. BMJ Open, 2019, 9, e028139.	1.9	126
14	Etiology and clinical presentation of birth defects: population based study. BMJ: British Medical Journal, 2017, 357, j2249.	2.3	125
15	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
16	Congenital Heart Defects, Maternal Febrile Illness, and Multivitamin Use: A Population-Based Study. Epidemiology, 2001, 12, 485-490.	2.7	112
17	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
18	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

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19	Acetaminophen Use in Pregnancy and Risk of Birth Defects. <i>Obstetrics and Gynecology</i> , 2010, 115, 109-115.	2.4	94
20	Maternal Fever, Multivitamin Use, and Selected Birth Defects: Evidence of Interaction?. <i>Epidemiology</i> , 2002, 13, 485-488.	2.7	91
21	Trends of selected malformations in relation to folic acid recommendations and fortification: An international assessment. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2006, 76, 693-705.	1.6	87
22	Biallelic Variants in UBA5 Reveal that Disruption of the UFM1 Cascade Can Result in Early-Onset Encephalopathy. <i>American Journal of Human Genetics</i> , 2016, 99, 695-703.	6.2	87
23	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. <i>American Journal of Human Genetics</i> , 2018, 103, 666-678.	6.2	87
24	Specific birth defects in pregnancies of women with diabetes: National Birth Defects Prevention Study, 1997â€“2011. <i>American Journal of Obstetrics and Gynecology</i> , 2020, 222, 176.e1-176.e11.	1.3	84
25	Maternal Exposure to Criteria Air Pollutants and Congenital Heart Defects in Offspring: Results from the National Birth Defects Prevention Study. <i>Environmental Health Perspectives</i> , 2014, 122, 863-872.	6.0	82
26	Maternal use of bupropion and risk for congenital heart defects. <i>American Journal of Obstetrics and Gynecology</i> , 2010, 203, 52.e1-52.e6.	1.3	78
27	Lack of periconceptional vitamins or supplements that contain folic acid and diabetes mellitusâ€“associated birth defects. <i>American Journal of Obstetrics and Gynecology</i> , 2012, 206, 218.e1-218.e13.	1.3	78
28	Cancer Risk in Children and Adolescents with Birth Defects: A Population-Based Cohort Study. <i>PLoS ONE</i> , 2013, 8, e69077.	2.5	67
29	Epidemiology and Prognosis of Congenital Diaphragmatic Hernia: A Populationâ€“Based Cohort Study in Utah. <i>Birth Defects Research</i> , 2017, 109, 1451-1459.	1.5	66
30	Associations between maternal periconceptional exposure to secondhand tobacco smoke and major birth defects. <i>American Journal of Obstetrics and Gynecology</i> , 2016, 215, 613.e1-613.e11.	1.3	51
31	Osteoporosis and skeletal dysplasia caused by pathogenic variants in SGMS2. <i>JCI Insight</i> , 2019, 4, .	5.0	47
32	Analysis of selected maternal exposures and nonâ€“syndromic atrioventricular septal defects in the National Birth Defects Prevention Study, 1997â€“2005.. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 2447-2455.	1.2	44
33	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. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2012, 94, 970-983.	1.6	43
34	Projected Changes in Maternal Heat Exposure During Early Pregnancy and the Associated Congenital Heart Defect Burden in the United States. <i>Journal of the American Heart Association</i> , 2019, 8, e010995.	3.7	41
35	SLC35A2â€“CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. <i>Human Mutation</i> , 2019, 40, 908-925.	2.5	39
36	Fostering International Collaboration in Birth Defects Research and Prevention: A Perspective From the International Clearinghouse for Birth Defects Surveillance and Research. <i>American Journal of Public Health</i> , 2006, 96, 774-780.	2.7	34

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37	How valid are the rates of Down syndrome internationally? Findings from the International Clearinghouse for Birth Defects Surveillance and Research. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 1670-1680.	1.2	34
38	Targeted gene panel sequencing for the rapid diagnosis of acutely ill infants. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e00796.	1.2	34
39	An evolutionary and developmental biology approach to gastroschisis. <i>Birth Defects Research</i> , 2019, 111, 294-311.	1.5	34
40	Surveillance of adverse fetal effects of medications (SAFE-Med): Findings from the International Clearinghouse of Birth Defects Surveillance and Research. <i>Reproductive Toxicology</i> , 2010, 29, 433-442.	2.9	33
41	Turner Syndrome in Girls Presenting with Coarctation of the Aorta. <i>Journal of Pediatrics</i> , 2015, 167, 1062-1066.	1.8	33
42	Effect of dietary lysine restriction and arginine supplementation in two patients with pyridoxine-dependent epilepsy. <i>Molecular Genetics and Metabolism</i> , 2016, 118, 167-172.	1.1	32
43	Lower rate of selected congenital heart defects with better maternal diet quality: a population-based study. <i>Archives of Disease in Childhood: Fetal and Neonatal Edition</i> , 2016, 101, 43-49.	2.8	32
44	Congenital heart defects after maternal fever. <i>American Journal of Obstetrics and Gynecology</i> , 2014, 210, 359.e1-359.e11.	1.3	29
45	Maternal periconceptional alcohol consumption and congenital heart defects. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2015, 103, 617-629.	1.6	27
46	Diagnosis, Treatment, and Clinical Outcome of Patients with Mitochondrial Trifunctional Protein/Long-Chain 3-Hydroxy Acyl-CoA Dehydrogenase Deficiency. <i>JIMD Reports</i> , 2016, 31, 63-71.	1.5	25
47	Is gastroschisis truly a sporadic defect? Familial cases of gastroschisis in Utah, 1997 to 2008. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2011, 91, 873-878.	1.6	24
48	Databases for Congenital Heart Defect Public Health Studies Across the Lifespan. <i>Journal of the American Heart Association</i> , 2016, 5, .	3.7	24
49	Maternal Exposure to Nitrogen Dioxide, Intake of Methyl Nutrients, and Congenital Heart Defects in Offspring. <i>American Journal of Epidemiology</i> , 2017, 186, 719-729.	3.4	24
50	A genome-wide association study implicates the BMP7 locus as a risk factor for nonsyndromic metopic craniosynostosis. <i>Human Genetics</i> , 2020, 139, 1077-1090.	3.8	24
51	Maternal Butalbital Use and Selected Defects in the National Birth Defects Prevention Study. <i>Headache</i> , 2014, 54, 54-66.	3.9	23
52	Loss-of-Function Mutations in ELMO2 Cause Intraosseous Vascular Malformation by Impeding RAC1 Signaling. <i>American Journal of Human Genetics</i> , 2016, 99, 299-317.	6.2	23
53	Developing a public health-tracking system for follow-up of newborn screening metabolic conditions: a four-state pilot project structure and initial findings. <i>Genetics in Medicine</i> , 2014, 16, 484-490.	2.4	22
54	Early Childhood Inpatient Costs of Critical Congenital Heart Disease. <i>Journal of Pediatrics</i> , 2018, 203, 371-379.e7.	1.8	22

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55	Expanding the genetic and clinical spectrum of the NONO-associated X-linked intellectual disability syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 792-796.	1.2	21
56	Clinical and biochemical outcomes of patients with medium-chain acyl-CoA dehydrogenase deficiency. <i>Molecular Genetics and Metabolism</i> , 2020, 129, 13-19.	1.1	21
57	Developing a research and public health agenda for gastroschisis: How do we bridge the gap between what is known and what is not?. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2008, 148C, 155-161.	1.6	20
58	Bayesian multinomial probit modeling of daily windows of susceptibility for maternal PM _{2.5} exposure and congenital heart defects. <i>Statistics in Medicine</i> , 2016, 35, 2786-2801.	1.6	19
59	Clinical presentation and survival in a population-based cohort of infants with gastroschisis in Utah, 1997-2011. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 306-315.	1.2	19
60	Triple surveillance: a proposal for an integrated strategy to support and accelerate birth defect prevention. <i>Annals of the New York Academy of Sciences</i> , 2018, 1414, 126-136.	3.8	19
61	Better Diet Quality before Pregnancy Is Associated with Reduced Risk of Gastroschisis in Hispanic Women. <i>Journal of Nutrition</i> , 2014, 144, 1781-1786.	2.9	17
62	Does Maternal Exposure to Secondhand Tobacco Smoke During Pregnancy Increase the Risk for Preterm or Small-for-Gestational Age Birth?. <i>Maternal and Child Health Journal</i> , 2018, 22, 1418-1429.	1.5	17
63	Case Definitions for Conditions Identified by Newborn Screening Public Health Surveillance. <i>International Journal of Neonatal Screening</i> , 2018, 4, 16.	3.2	17
64	Congenital anomalies and associated risk factors in a Saudi population: a cohort study from pregnancy to age 2 years. <i>BMJ Open</i> , 2019, 9, e026351.	1.9	17
65	Newborn screening for critical congenital heart disease: Essential public health roles for birth defects monitoring programs. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2012, 94, 965-969.	1.6	16
66	Risk of gastroschisis with maternal genitourinary infections: the US National birth defects prevention study 1997-2011. <i>BMJ Open</i> , 2019, 9, e026297.	1.9	16
67	Associations between PM2.5 and risk of preterm birth among liveborn infants. <i>Annals of Epidemiology</i> , 2019, 39, 46-53.e2.	1.9	15
68	Identification of fibrinogen as a natural inhibitor of MMP-2. <i>Scientific Reports</i> , 2019, 9, 4340.	3.3	15
69	Maternal intake of vitamin E and birth defects, national birth defects prevention study, 1997 to 2005. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2014, 100, 647-657.	1.6	14
70	Risk factors for Dandy-Walker malformation: A population-based assessment. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2009-2016.	1.2	14
71	Elevated body mass index and decreased diet quality among women and risk of birth defects in their offspring. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2016, 106, 164-171.	1.6	14
72	Mild orotic aciduria in <i>UMPS</i> heterozygotes: a metabolic finding without clinical consequences. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 423-431.	3.6	14

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73	Maternal exposure to outdoor air pollution and congenital limb deficiencies in the National Birth Defects Prevention Study. <i>Environmental Research</i> , 2019, 179, 108716.	7.5	14
74	Reflections on the etiology of structural birth defects: Established teratogens and risk factors. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2015, 103, 652-655.	1.6	13
75	Brief Report: Pediatrician Perspectives Regarding Genetic Evaluations of Children with Autism Spectrum Disorder. <i>Journal of Autism and Developmental Disorders</i> , 2019, 49, 794-808.	2.7	13
76	Join World Birth Defects Day. <i>Pediatric Research</i> , 2019, 86, 3-4.	2.3	12
77	Maternal Smoking and Congenital Heart Defects, National Birth Defects Prevention Study, 1997-2011. <i>Journal of Pediatrics</i> , 2022, 240, 79-86.e1.	1.8	12
78	Building capacity for birth defects surveillance in Africa: Implementation of an intermediate birth defects surveillance workshop. <i>Journal of Global Health Perspectives</i> , 2015, 2015, .	0.3	11
79	Risk factors associated with the development of double-inlet ventricle congenital heart disease. <i>Birth Defects Research</i> , 2019, 111, 640-648.	1.5	10
80	Evaluating cost and resource use associated with pulse oximetry screening for critical congenital heart disease: Empiric estimates and sources of variation. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2015, 103, 962-971.	1.6	9
81	Modification of the association between diabetes and birth defects by obesity, National Birth Defects Prevention Study, 1997-2011. <i>Birth Defects Research</i> , 2021, 113, 1084-1097.	1.5	9
82	Value of sharing and networking among birth defects surveillance programs: an ICBDSR perspective. <i>Journal of Community Genetics</i> , 2018, 9, 411-415.	1.2	8
83	From cause to care: Triple surveillance for better outcomes in birth defects and rare diseases. <i>European Journal of Medical Genetics</i> , 2018, 61, 551-555.	1.3	8
84	Survival of infants born with esophageal atresia among 24 international birth defects surveillance programs. <i>Birth Defects Research</i> , 2021, 113, 945-957.	1.5	8
85	Flavors in Gene-Environment Interactions. <i>Epidemiology</i> , 2007, 18, 431-432.	2.7	6
86	Acardia: Epidemiologic findings and literature review from the International Clearinghouse for Birth Defects Surveillance and Research. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2011, 157, 262-273.	1.6	6
87	Global birth defects app: An innovative tool for describing and coding congenital anomalies at birth in low resource settings. <i>Birth Defects Research</i> , 2021, 113, 1057-1073.	1.5	6
88	Costs, mortality, and hospital usage in relation to prenatal diagnosis in d-transposition of the great arteries. <i>Birth Defects Research</i> , 2017, 109, 262-270.	1.5	5
89	Delineation of the 9q31 deletion syndrome: Genomic microarray characterization of two patients with overlapping deletions. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2901-2906.	1.2	4
90	From cause to care: Can a triple approach to better population data improve the global outlook of congenital heart disease?. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2020, 184, 23-35.	1.6	4

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91	Patterns of multiple congenital anomalies in the National Birth Defect Prevention Study: Challenges and insights. <i>Birth Defects Research</i> , 2023, 115, 43-55.	1.5	4
92	Intracranial Calcifications in Young Children. <i>Seminars in Pediatric Neurology</i> , 2018, 26, 135-139.	2.0	3
93	Chondrodysplasia and growth failure in children after early hematopoietic stem cell transplantation for non-oncologic disorders. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 517-527.	1.2	3
94	Application of quality indicators to data from the National Network of Congenital Anomalies of Argentina. <i>Birth Defects Research</i> , 2019, 111, 333-340.	1.5	2
95	A proposal for the systematic assessment of data quality indicators in birth defects surveillance. <i>Birth Defects Research</i> , 2019, 111, 324-332.	1.5	2
96	Paternal genetic variants and risk of obstructive heart defects: A parent-of-origin approach. <i>PLoS Genetics</i> , 2021, 17, e1009413.	3.5	2
97	Comparative Serum Analyses Identify Cytokines and Hormones Commonly Dysregulated as Well as Implicated in Promoting Osteolysis in MMP-2-Deficient Mice and Children. <i>Frontiers in Physiology</i> , 2020, 11, 568718.	2.8	1
98	Skeletal dysplasias in art and antiquities: A cultural journey through genes, environment, and chance. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2021, 187, 199-212.	1.6	1
99	Maternal plasma and erythrocyte folate levels and risk of oral clefts in Utah. <i>FASEB Journal</i> , 2007, 21, .	0.5	0
100	Development and Utility of a Birth Defects Surveillance Toolkit. <i>Journal of Global Health Perspectives</i> , 2018, 0, .	0.3	0