

Sophie Collardeau-Frachon

List of Publications by Year in descending order

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Version: 2024-02-01

61
papers

1,523
citations

393982

19
h-index

344852

36
g-index

68
all docs

68
docs citations

68
times ranked

2817
citing authors

#	ARTICLE	IF	CITATIONS
1	Vascular Development and Differentiation During Human Liver Organogenesis. <i>Anatomical Record</i> , 2008, 291, 614-627.	0.8	140
2	Placental Tissue Destruction and Insufficiency From COVID-19 Causes Stillbirth and Neonatal Death From Hypoxic-Ischemic Injury. <i>Archives of Pathology and Laboratory Medicine</i> , 2022, 146, 660-676.	1.2	127
3	Homozygous MTTP and APOB mutations may lead to hepatic steatosis and fibrosis despite metabolic differences in congenital hypocholesterolemia. <i>Journal of Hepatology</i> , 2014, 61, 891-902.	1.8	116
4	Loss of β 1 Soluble Guanylate Cyclase, the Major Nitric Oxide Receptor, Leads to Moyamoya and Achalasia. <i>American Journal of Human Genetics</i> , 2014, 94, 385-394.	2.6	95
5	Mutations in CECR1 associated with a neutrophil signature in peripheral blood. <i>Pediatric Rheumatology</i> , 2014, 12, 44.	0.9	88
6	Novel NEK8 Mutations Cause Severe Syndromic Renal Cystic Dysplasia through YAP Dysregulation. <i>PLoS Genetics</i> , 2016, 12, e1005894.	1.5	77
7	Defects in t6A tRNA modification due to GON7 and YRDC mutations lead to Galloway-Mowat syndrome. <i>Nature Communications</i> , 2019, 10, 3967.	5.8	66
8	DUX4 and DUX4 downstream target genes are expressed in fetal FSHD muscles. <i>Human Molecular Genetics</i> , 2014, 23, 171-181.	1.4	61
9	<i>DCDC2</i> Mutations Cause Neonatal Sclerosing Cholangitis. <i>Human Mutation</i> , 2016, 37, 1025-1029.	1.1	56
10	Placental lesions and SARS-Cov-2 infection: Diffuse placenta damage associated to poor fetal outcome. <i>Placenta</i> , 2021, 112, 97-104.	0.7	55
11	Neonatal Hemochromatosis: Diagnostic Work-Up Based on a Series of Cases of Fetal Death and Neonatal Liver Failure. <i>Journal of Pediatrics</i> , 2015, 166, 66-73.	0.9	38
12	The protein kinase PERK/EIF2AK3 regulates proinsulin processing not via protein synthesis but by controlling endoplasmic reticulum chaperones. <i>Journal of Biological Chemistry</i> , 2018, 293, 5134-5149.	1.6	33
13	French Retrospective Multicentric Study of Neonatal Hemochromatosis: Importance of Autopsy and Autoimmune Maternal Manifestations. <i>Pediatric and Developmental Pathology</i> , 2012, 15, 450-470.	0.5	30
14	Fetal anomalies associated with <i>HNF1B</i> mutations: report of 20 autopsy cases. <i>Prenatal Diagnosis</i> , 2016, 36, 744-751.	1.1	28
15	Antenatal manifestations of inborn errors of metabolism: biological diagnosis. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 611-624.	1.7	27
16	Gut Inflammation in Mice Triggers Proliferation and Function of Mucosal Foxp3+Regulatory T Cells but Impairs Their Conversion from CD4+T Cells. <i>Journal of Crohn's and Colitis</i> , 2017, 11, 105-117.	0.6	24
17	IMMUNOHISTOCHEMICAL EXPRESSION OF P57 IN PLACENTAL VASCULAR PROLIFERATIVE DISORDERS OF PRETERM AND TERM PLACENTAS. <i>Fetal and Pediatric Pathology</i> , 2009, 28, 9-23.	0.4	23
18	ALG3 β CDG: Report of two siblings with antenatal features carrying homozygous p.Gly96Arg mutation. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2748-2754.	0.7	21

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19	Deleterious impact of C3d-binding donor-specific anti-HLA antibodies after pediatric liver transplantation. <i>Transplant Immunology</i> , 2017, 45, 8-14.	0.6	21
20	EBV virus-associated smooth muscle tumors in a composite tissue allograft and a pediatric liver transplant recipient. <i>Transplant Infectious Disease</i> , 2013, 15, E182-6.	0.7	20
21	Assisted Reproductive Technologies and imprinting disorders: Results of a study from a French congenital malformations registry. <i>European Journal of Medical Genetics</i> , 2018, 61, 518-523.	0.7	20
22	A novel morphological approach to gonads in disorders of sex development. <i>Modern Pathology</i> , 2016, 29, 1399-1414.	2.9	19
23	Second Malignant Neoplasms Following Childhood Cancer: A Study of a Recent Cohort (1987-2004) from the Childhood Cancer Registry of The Rhône-Alpes Region (ARCERRA) in France. <i>Pediatric Hematology and Oncology</i> , 2011, 28, 364-379.	0.3	17
24	Liver transplantation of partial grafts after ex situ splitting during hypothermic oxygenated perfusion: The HOPE Split pilot study. <i>Liver Transplantation</i> , 2022, 28, 1576-1587.	1.3	17
25	Primary Desmoplastic Small round Cell Tumor of the Kidney: A Case Report in a 14-Year-Old Girl with Molecular Confirmation. <i>Pediatric and Developmental Pathology</i> , 2007, 10, 320-324.	0.5	16
26	Long-term treatment reduction and steroids withdrawal in children with autoimmune hepatitis: a single centre experience on 55 children. <i>European Journal of Gastroenterology and Hepatology</i> , 2009, 21, 1413-1418.	0.8	15
27	Transient Neonatal Liver Disease After Maternal Antenatal Intravenous Ig Infusions in Gestational Alloimmune Liver Disease Associated With Neonatal Haemochromatosis. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2014, 59, 629-635.	0.9	15
28	Small Cell Carcinoma of the Ovary, Hypercalcemic Type: Report of a Bilateral Case in a Teenager Associated with SMARCA4 Germline Mutation. <i>Pediatric and Developmental Pathology</i> , 2016, 19, 56-60.	0.5	15
29	Relevance of C5b9 immunostaining in the diagnosis of neonatal hemochromatosis. <i>Pediatric Research</i> , 2017, 81, 712-721.	1.1	15
30	Microscopic and ultrastructural features in Wolcott-Rallison syndrome, a permanent neonatal diabetes mellitus: about two autopsy cases. <i>Pediatric Diabetes</i> , 2015, 16, 510-520.	1.2	13
31	Radiographic features of the skeleton in disorders of post-squalene cholesterol biosynthesis. <i>Pediatric Radiology</i> , 2015, 45, 965-976.	1.1	12
32	Antenatal manifestations of inborn errors of metabolism: prenatal imaging findings. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 103-112.	1.7	12
33	Prenatal imaging features suggestive of liver gestational allo immune disease. <i>Journal of Gynecology Obstetrics and Human Reproduction</i> , 2019, 48, 61-64.	0.6	12
34	Long-term disease course in a patient with severe neonatal IPEX syndrome. <i>Clinics and Research in Hepatology and Gastroenterology</i> , 2015, 39, e43-e47.	0.7	11
35	Antenatal manifestations of inborn errors of metabolism: autopsy findings suggestive of a metabolic disorder. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 597-610.	1.7	11
36	Childhood/adult-onset lysosomal acid lipase deficiency: A serious metabolic and vascular phenotype beyond liver disease: four new pediatric cases. <i>Journal of Clinical Lipidology</i> , 2017, 11, 167-177.e3.	0.6	10

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37	Normal serum ApoB48 and red cells vitamin E concentrations after supplementation in a novel compound heterozygous case of abetalipoproteinemia. <i>Atherosclerosis</i> , 2019, 284, 75-82.	0.4	10
38	Unexpected diagnosis of cystic fibrosis at liver biopsy: a report of four pediatric cases. <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , 2007, 451, 57-64.	1.4	9
39	Lipid profile and cardiovascular risk factors in pediatric liver transplant recipients. <i>Pediatric Transplantation</i> , 2016, 20, 241-248.	0.5	9
40	A botryoid rhabdomyosarcoma diagnosed as a choledochal cyst. <i>Pediatric Blood and Cancer</i> , 2013, 60, 2089-2090.	0.8	7
41	Prenatal diagnosis of osteopathia striata with cranial sclerosis. <i>Prenatal Diagnosis</i> , 2015, 35, 302-304.	1.1	7
42	In utero ultrasound diagnosis of corpus callosum agenesis leading to the identification of orofacioidigital type 1 syndrome in female fetuses. <i>Birth Defects Research</i> , 2018, 110, 382-389.	0.8	7
43	NLRC4 GOF Mutations, a Challenging Diagnosis from Neonatal Age to Adulthood. <i>Journal of Clinical Medicine</i> , 2021, 10, 4369.	1.0	7
44	Escherichia coli-associated hemolytic uremic syndrome and severe chronic hepatocellular cholestasis: complication or side effect of eculizumab?. <i>Pediatric Nephrology</i> , 2019, 34, 1289-1293.	0.9	6
45	Management and treatment of a sialoblastoma of the submandibular gland in a neonate: Report of one case. <i>International Journal of Pediatric Otorhinolaryngology Extra</i> , 2011, 6, 168-171.	0.1	5
46	Fetal cerebral hemorrhage due to Xâ€linked <i>GATA1</i> gene mutation. <i>Prenatal Diagnosis</i> , 2018, 38, 772-778.	1.1	5
47	Kimura Disease Mimicking an Aneurysm of the Radial Artery. <i>Journal of Pediatrics</i> , 2015, 167, 1166-1166.e2.	0.9	4
48	Pulmonary Infantile Hemangioma Mimicking a Congenital Cystic Adenomatoid Malformation. <i>Pediatric and Developmental Pathology</i> , 2019, 22, 480-485.	0.5	4
49	Drug-Induced Fulminant Hepatitis in a Child Treated for Latent Multidrug-Resistant Tuberculosis With Dual Therapy Combining Pyrazinamide and Levofloxacin. <i>Pediatric Infectious Disease Journal</i> , 2019, 38, 1025-1026.	1.1	4
50	Children with eosinophilic esophagitis in real life: 10 yearsâ€™ experience with a focus on allergic management. <i>Allergologia Et Immunopathologia</i> , 2020, 48, 244-250.	1.0	4
51	Hepatocyte proteomes reveal the role of protein disulfide isomerase 4 in alpha 1-antitrypsin deficiency. <i>JHEP Reports</i> , 2021, 3, 100297.	2.6	4
52	It sounds like a relapsing polychondritis. <i>Lancet Infectious Diseases</i> , The, 2013, 13, 638.	4.6	3
53	Invasive Hepatobiliary <i>Trichosporon asahii</i> Infection in a Child With Autoimmune Cholangitis. <i>Journal of the Pediatric Infectious Diseases Society</i> , 2019, 8, 574-577.	0.6	2
54	Sarcoma Occurring at the Site of Growth Hormone Therapy. <i>Journal of Pediatric Hematology/Oncology</i> , 2020, 42, 335-335.	0.3	2

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55	Gastrointestinal lymphoid pseudotumoral hyperplasia: report of four pediatric cases. <i>Endoscopy</i> , 2008, 40, E267-E268.	1.0	1
56	Management and treatment of a sialoblastoma of the submandibular gland in a neonate. <i>International Journal of Pediatric Otorhinolaryngology Extra</i> , 2011, 6, 9-12.	0.1	1
57	A quest for Q fever. <i>Lancet, The</i> , 2019, 394, 419.	6.3	1
58	Loss of β 1 Soluble Guanylate Cyclase, the Major Nitric Oxide Receptor, Leads to Moyamoya and Achalasia. <i>American Journal of Human Genetics</i> , 2014, 94, 642.	2.6	0
59	Donor-Related Coccidioidomycosis Transmitted Through Left-Liver Transplant to a Child Recipient in a Nonendemic Area. <i>Pediatric Infectious Disease Journal</i> , 2017, 36, 805-808.	1.1	0
60	Hemorrhagic and necrotic adenoma associated with a congenital portosystemic shunt. <i>Digestive and Liver Disease</i> , 2020, 52, 920-921.	0.4	0
61	Expanding the Phenotypic Spectrum of Encephalocraniocutaneous Lipomatosis: About a Prenatal Case With Complete Autopsy. <i>Pediatric and Developmental Pathology</i> , 2021, , 109352662110408.	0.5	0