Pierre-Simon Jouk

List of Publications by Year
in descending order

111	3,900	159585 30	144013 57
papers	citations	h-index	g-index

128	128	128	5660
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Phenotypic spectrum and genomics of undiagnosed arthrogryposis multiplex congenita. Journal of Medical Genetics, 2022, 59, 559-567.	3.2	25
2	PIGNencephalopathy: Characterizing the epileptology. Epilepsia, 2022, 63, 974-991.	5.1	4
3	The Myosin Myocardial Mesh Interpreted as a Biological Analogous of Nematic Chiral Liquid Crystals. Journal of Cardiovascular Development and Disease, 2021, 8, 179.	1.6	3
4	Diagnostic workup in children with arthrogryposis: description of practices from a single reference centre, comparison with literature and suggestion of recommendations. Journal of Medical Genetics, 2021, , jmedgenet-2021-107823.	3.2	1
5	Longitudinal Study by Two-Dimensional Speckle-Tracking Echocardiography of the Left Ventricle Rotational Mechanics during Postnatal Adaptation in Healthy Newborns. Journal of the American Society of Echocardiography, 2020, 33, 252-254.	2.8	7
6	High Activation of the AKT Pathway in Human Multicystic Renal Dysplasia. Pathobiology, 2020, 87, 302-310.	3.8	4
7	Nextâ€generation sequencing in a series of 80 fetuses with complex cardiac malformations and/or heterotaxy. Human Mutation, 2020, 41, 2167-2178.	2.5	21
8	Prenatal Diagnosis of Aorto-Left Ventricular Tunnel With Dysplastic Bicuspid Aortic Valve: From Fetal Cardiac Failure to Favorable Outcome. Frontiers in Pediatrics, 2020, 8, 69.	1.9	4
9	TAR syndrome: Clinical and molecular characterization of a cohort of 26 patients and description of novel noncoding variants of RBM8A. Human Mutation, 2020, 41, 1220-1225.	2.5	17
10	Excess of de novo variants in genes involved in chromatin remodelling in patients with marfanoid habitus and intellectual disability. Journal of Medical Genetics, 2020, 57, 466-474.	3.2	7
11	Polarized Light Imaging of the Myoarchitecture in Tetralogy of Fallot in the Perinatal Period. Frontiers in Pediatrics, 2020, 8, 503054.	1.9	2
12	Major intra-familial phenotypic heterogeneity and incomplete penetrance due to a CACNA1A pathogenic variant. European Journal of Medical Genetics, 2019, 62, 103530.	1.3	34
13	The diagnostic workup in a patient with AMC: Overview of the clinical evaluation and paraclinical analyses with review of the literature. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 337-344.	1.6	15
14	Author response: Disability in adults with arthrogryposis is severe, partly invisible, and varies by genotype. Neurology, 2019, 92, 636.1-636.	1.1	0
15	Dysregulations of sonic hedgehog signaling in MED12 â€related Xâ€linked intellectual disability disorders. Molecular Genetics & Genomic Medicine, 2019, 7, e00569.	1.2	14
16	Xq22.3q23 microdeletion harboring TMEM164 and AMMECR1 genes: Two case reports confirming a recognizable phenotype with short stature, midface hypoplasia, intellectual delay, and elliptocytosis. American Journal of Medical Genetics, Part A, 2019, 179, 650-654.	1.2	2
17	Further delineation of theMECP2duplication syndrome phenotype in 59 French male patients, with a particular focus on morphological and neurological features. Journal of Medical Genetics, 2018, 55, 359-371.	3.2	45
18	Disability in adults with arthrogryposis is severe, partly invisible, and varies by genotype. Neurology, 2018, 90, e1596-e1604.	1.1	25

#	Article	IF	Citations
19	Mutations in CFAP43 and CFAP44 cause male infertility and flagellum defects in Trypanosoma and human. Nature Communications, 2018, 9, 686.	12.8	173
20	CHARGE syndrome: a recurrent hotspot of mutations in CHD7 IVS25 analyzed by bioinformatic tools and minigene assays. European Journal of Human Genetics, 2018, 26, 287-292.	2.8	7
21	Postnatal myocardium remodelling generates inhomogeneity in the architecture of the ventricular mass. Surgical and Radiologic Anatomy, 2018, 40, 75-83.	1.2	11
22	An analytical fiber ODF reconstruction in 3D polarized light imaging. , 2018, , .		4
23	Prenatally diagnosed periventricular nodular heterotopia: Further delineation of the imaging phenotype and outcome. European Journal of Medical Genetics, 2018, 61, 773-782.	1.3	8
24	Quantitative comparison of human myocardial fiber orientations derived from DTI and polarized light imaging. Physics in Medicine and Biology, 2018, 63, 215003.	3.0	14
25	Genomic duplication in the 19q13.42 imprinted region identified as a new genetic cause of intrauterine growth restriction. Clinical Genetics, 2018, 94, 575-580.	2.0	12
26	Clinical and molecular cytogenetic characterization of four unrelated patients carrying 2p14 microdeletions. American Journal of Medical Genetics, Part A, 2017, 173, 2268-2274.	1.2	3
27	MED12-related XLID disorders are dose-dependent of immediate early genes (IEGs) expression. Human Molecular Genetics, 2017, 26, 2062-2075.	2.9	24
28	PBX1haploinsufficiency leads to syndromic congenital anomalies of the kidney and urinary tract (CAKUT) in humans. Journal of Medical Genetics, 2017, 54, 502-510.	3.2	46
29	Microduplication of the ARID1A gene causes intellectual disability with recognizable syndromic features. Genetics in Medicine, 2017, 19, 701-710.	2.4	13
30	Fetal hypothyroidism induced by maternal anti-TSH receptor blocking antibodies and complicated by polyhydramnios despite the absence of goiter. Treatment by intra-amniotic injections of levothyroxine. Annales D'Endocrinologie, 2017, 78, 61-64.	1.4	2
31	Biallelic mutation of UNC50, encoding a protein involved in AChR trafficking, is responsible for arthrogryposis. Human Molecular Genetics, 2017, 26, 3989-3994.	2.9	8
32	Clinicoâ€molecular analysis of eleven patients with Hermanskyâ€“Pudlak type 5 syndrome, a mild form of <scp>HPS</scp>. Pigment Cell and Melanoma Research, 2017, 30, 563-570.	3.3	13
33	Phenotype and genotype analysis of a French cohort of 119 patients with CHARGE syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 417-430.	1.6	65
34	Xq28 duplication including <scp>MECP2</scp> in six unreported affected females: what can we learn for diagnosis and genetic counselling?. Clinical Genetics, 2017, 91, 576-588.	2.0	17
35	Microdeletion del(22)(q12.1) excluding the MN1 gene in a patient with craniofacial anomalies. American Journal of Medical Genetics, Part A, 2016, 170, 498-503.	1.2	6
36	Study of myocardial cell inhomogeneity of the human heart: Simulation and validation using polarized light imaging. Medical Physics, 2016, 43, 2273-2282.	3.0	11

#	Article	IF	Citations
37	Large national series of patients with Xq28 duplication involving MECP2: Delineation of brain MRI abnormalities in 30 affected patients. American Journal of Medical Genetics, Part A, 2016, 170, 116-129.	1.2	19
38	9q33.3q34.11 microdeletion: new contiguous gene syndrome encompassing STXBP1, LMX1B and ENG genes assessed using reverse phenotyping. European Journal of Human Genetics, 2016, 24, 830-837.	2.8	13
39	Low but Increasing Prevalence of Autism Spectrum Disorders in a French Area from Register-Based Data. Journal of Autism and Developmental Disorders, 2015, 45, 3255-3261.	2.7	39
40	13q31.1 microdeletion: A prenatal case report with macrocephaly and macroglossia. European Journal of Medical Genetics, 2015, 58, 526-530.	1.3	6
41	Array-CGH in children with mild intellectual disability: a population-based study. European Journal of Pediatrics, 2015, 174, 75-83.	2.7	16
42	Very High-Resolution Imaging of Post-Mortem Human Cardiac Tissue Using X-Ray Phase Contrast Tomography. Lecture Notes in Computer Science, 2015, , 172-179.	1.3	10
43	Currarino Syndrome and HPE Microform Associated with a 2.7-Mb Deletion in 7q36.3 Excluding SHH Gene. Molecular Syndromology, 2014, 5, 25-31.	0.8	9
44	Mutations in CNTNAP1 and ADCY6 are responsible for severe arthrogryposis multiplex congenita with axoglial defects. Human Molecular Genetics, 2014, 23, 2279-2289.	2.9	98
45	Mutations in DNAH1, which Encodes an Inner Arm Heavy Chain Dynein, Lead to Male Infertility from Multiple Morphological Abnormalities of the Sperm Flagella. American Journal of Human Genetics, 2014, 94, 95-104.	6.2	328
46	Split hand/foot malformation with longâ€bone deficiency and <scp>BHLHA9</scp> duplication: report of 13 new families. Clinical Genetics, 2014, 85, 464-469.	2.0	20
47	Maternal complex chromosomal rearrangement leads to TCF12 microdeletion in a patient presenting with coronal craniosynostosis and intellectual disability. American Journal of Medical Genetics, Part A, 2014, 164, 1530-1536.	1.2	11
48	SFCP CO-13 - Contribution Ã lâ€™Ã©tude anatomique et embryologique des kystes congÃ©nitaux dÃ©rivÃ©s du proentÃ©ron. Archives De Pediatrie, 2014, 21, 391.	1.0	0
49	7p22.3 microdeletion disrupting SNX8 in a patient presenting with intellectual disability but no tetralogy of Fallot. American Journal of Medical Genetics, Part A, 2014, 164, 2133-2135.	1.2	7
50	Prevalence and characteristics of children with mild intellectual disability in a <scp>F</scp>rench county. Journal of Intellectual Disability Research, 2014, 58, 591-602.	2.0	22
51	Delineation of the 3p14.1p13 microdeletion associated with syndromic distal limb contractures. American Journal of Medical Genetics, Part A, 2014, 164, 3027-3034.	1.2	6
52	Complete agenesis of major salivary glands. International Journal of Pediatric Otorhinolaryngology, 2013, 77, 1782-1785.	1.0	14
53	Systematic molecular and cytogenetic screening of 100 patients with marfanoid syndromes and intellectual disability. Clinical Genetics, 2013, 84, 507-521.	2.0	23
54	Fine Characterisation of a Recombination Hotspot at the DPY19L2 Locus and Resolution of the Paradoxical Excess of Duplications over Deletions in the General Population. PLoS Genetics, 2013, 9, e1003363.	3.5	25

#	Article	IF	Citations
55	The neuronal endopeptidase ECEL1 is associated with a distinct form of recessive distal arthrogryposis. Human Molecular Genetics, 2013, 22, 1483-1492.	2.9	66
56	Interphase fluorescent in situ hybridization detection of the 7q11.23 chromosomal inversion in a clinical laboratory: automated versus manual scoring. Clinical Chemistry and Laboratory Medicine, 2013, 51, e41-4.	2.3	1
57	190â€‰kb duplication in 1p36.11 including <scp>PIGV</scp> and <scp>ARID1A</scp> genes in a girl with intellectual disability and hexadactyly. Clinical Genetics, 2013, 84, 596-599.	2.0	2
58	Stillbirth classification in population-based data and role of fetal growth restriction: the example of RECODE. BMC Pregnancy and Childbirth, 2013, 13, 182.	2.4	21
59	Modeling of the Optical Behavior of Myocardial Fibers in Polarized Light Imaging. Lecture Notes in Computer Science, 2013, , 235-244.	1.3	1
60	Identification of a new recurrent Aurora kinase C mutation in both European and African men with macrozoospermia. Human Reproduction, 2012, 27, 3337-3346.	0.9	52
61	MLPA and sequence analysis of DPY19L2 reveals point mutations causing globozoospermia. Human Reproduction, 2012, 27, 2549-2558.	0.9	62
62	17p13.1 microduplication in a boy with Silverâ€“Russell syndrome features and intellectual disability. American Journal of Medical Genetics, Part A, 2012, 158A, 2564-2570.	1.2	14
63	Recurrent Dominant Mutations Affecting Two Adjacent Residues in the Motor Domain of the Monomeric Kinesin KIF22 Result in Skeletal Dysplasia and Joint Laxity. American Journal of Human Genetics, 2012, 90, 170.	6.2	0
64	Clinical and mutational spectrum in a cohort of 105 unrelated patients with dilated cardiomyopathy. European Journal of Medical Genetics, 2011, 54, e570-e575.	1.3	92
65	A Recurrent Deletion of DPY19L2 Causes Infertility in Man by Blocking Sperm Head Elongation and Acrosome Formation. American Journal of Human Genetics, 2011, 88, 351-361.	6.2	165
66	Recurrent Dominant Mutations Affecting Two Adjacent Residues in the Motor Domain of the Monomeric Kinesin KIF22 Result in Skeletal Dysplasia and Joint Laxity. American Journal of Human Genetics, 2011, 89, 767-772.	6.2	31
67	Association of TALS Developmental Disorder with Defect in Minor Splicing Component U4atac snRNA. Science, 2011, 332, 240-243.	12.6	195
68	Swallowed amniotic band presenting as a cystic mass of the fetal nasal cavity. Ultrasound in Obstetrics and Gynecology, 2010, 35, 246-247.	1.7	3
69	Paracentric inversion of chromosome 2 associated with cryptic duplication of 2q14 and deletion of 2q37 in a patient with autism. American Journal of Medical Genetics, Part A, 2010, 152A, 2346-2354.	1.2	17
70	Identification of new FOXP3 mutations and prenatal diagnosis of IPEX syndrome. Prenatal Diagnosis, 2010, 30, 1072-1078.	2.3	39
71	Renal insufficiency, a frequent complication with age in oralâ€‰facialâ€‰digital syndrome type I. Clinical Genetics, 2010, 77, 258-265.	2.0	29
72	Brachytelephalangic Chondrodysplasia Punctata: Prenatal Diagnosis and Postnatal Outcome. Fetal Diagnosis and Therapy, 2010, 28, 186-190.	1.4	17

#	Article	IF	Citations
73	Prevalence and spectrum of mutations in a cohort of 192 unrelated patients with hypertrophic cardiomyopathy. European Journal of Medical Genetics, 2010, 53, 261-267.	1.3	130
74	The Aurora Kinase C c.144delC mutation causes meiosis I arrest in men and is frequent in the North African population. Human Molecular Genetics, 2009, 18, 1301-1309.	2.9	97
75	Prenatal diagnosis of achondroplasia: new specific signs. Prenatal Diagnosis, 2009, 29, 697-702.	2.3	37
76	Absence of Î²-tropomyosin is a new cause of Escobar syndrome associated with nemaline myopathy. Neuromuscular Disorders, 2009, 19, 118-123.	0.6	58
77	Prenatal manifestation in a family affected by nevoid basal cell carcinoma syndrome. European Journal of Medical Genetics, 2008, 51, 472-478.	1.3	9
78	Rare diseases in disabled children: an epidemiological survey. Archives of Disease in Childhood, 2008, 93, 115-118.	1.9	25
79	Analysis of the fiber architecture of the heart by quantitative polarized light microscopy. Accuracy, limitations and contribution to the study of the fiber architecture of the ventricles during fetal and neonatal lifeâ˜†. European Journal of Cardio-thoracic Surgery, 2007, 31, 915-921.	1.4	49
80	C.P.1.13 A homozygous null mutation in TPM2 gene causes autosomal recessive nemaline myopathy associated with multiple pterygia. Neuromuscular Disorders, 2007, 17, 837-838.	0.6	2
81	Could ovarian choriocarcinoma be detected by maternal serum screening for Down syndrome?. Prenatal Diagnosis, 2007, 27, 682-684.	2.3	3
82	Homozygous mutation of AURKC yields large-headed polyploid spermatozoa and causes male infertility. Nature Genetics, 2007, 39, 661-665.	21.4	248
83	Isolation of microsatellite markers for the endangered Knysna seahorseHippocampus capensisand their use in the detection of a genetic bottleneck. Molecular Ecology Notes, 2007, 7, 638-640.	1.7	15
84	Quality control of an image-scoring method for nuchal translucency ultrasonography. American Journal of Obstetrics and Gynecology, 2007, 196, 272.e1-272.e5.	1.3	40
85	Very early prophylactic thyroid surgery for infants with a mutation of the RET proto-oncogene at codon 634: evaluation of the implementation of international guidelines for MEN type 2 in a single centre. Clinical Endocrinology, 2006, 65, 118-124.	2.4	15
86	Fetal lung volume in congenital diaphragmatic hernia. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2006, 91, F363-F364.	2.8	43
87	Trends, perinatal characteristics, and medical conditions in pervasive developmental disorders. Developmental Medicine and Child Neurology, 2006, 48, 896-900.	2.1	0
88	Trends, perinatal characteristics, and medical conditions in pervasive developmental disorders. Developmental Medicine and Child Neurology, 2006, 48, 896.	2.1	27
89	More on: asymptomatic thrombophilia-a family affair. Journal of Thrombosis and Haemostasis, 2005, 3, 1329-1330.	3.8	4
90	Perforated tubular duplication of the transverse colon: a rare cause of meconium peritonitis with prenatal diagnosis. Pediatric Surgery International, 2005, 21, 110-112.	1.4	16

#	Article	IF	Citations
91	Late event-related potentials and movement complexity in young adults with Down syndrome. Neurophysiologie Clinique, 2005, 35, 81-91.	2.2	16
92	A submicroscopic unbalanced subtelomeric translocation t(2p;10q) identified by fluorescence in situ hybridization: fetus with increased nuchal translucency and normal standard karyotype with later growth and developmental delay, rhombencephalosynapsis (RES). Annales De GÃ©nÃ©tique, 2004, 47, 405-417.	0.4	15
93	Trends in elective terminations of pregnancy between 1989 and 2000 in a French county (the IsÃ¨re). Prenatal Diagnosis, 2003, 23, 877-883.	2.3	46
94	Cerebral palsy and intrauterine growth in single births: European collaborative study. Lancet, The, 2003, 362, 1106-1111.	13.7	297
95	Disabilities and trends over time in a French county, 1980-91. Archives of Disease in Childhood, 2003, 88, 114-117.	1.9	30
96	Carbimazole-Related Gastroschisis. Annals of Pharmacotherapy, 2003, 37, 829-831.	1.9	17
97	A Model of the Structural and Functional Development of the Normal Human Fetal Left Ventricle Based on a Global Growth Law. Computer Methods in Biomechanics and Biomedical Engineering, 2002, 5, 113-126.	1.6	6
98	So-called â€˜cryptogenicâ€™ partial seizures resulting from a subtle cortical dysgenesis due to adoublecortin gene mutation. Seizure: the Journal of the British Epilepsy Association, 2002, 11, 273-277.	2.0	14
99	Diastrophic dwarfism and pregnancy. Lancet, The, 2001, 358, 1778.	13.7	9
100	A heterozygous endothelin 3 mutation in Waardenburg-Hirschsprung disease: is there a dosage effect of EDN3/EDNRB gene mutations on neurocristopathy phenotypes?. Journal of Medical Genetics, 2001, 38, 205-209.	3.2	50
101	Three-dimensional cartography of the pattern of the myofibres in the second trimester fetal human heart. Anatomy and Embryology, 2000, 202, 103-118.	1.5	97
102	Fibre Orientation in Human Fetal Heart and Ventricular Mechanics : A Small Perturbation. Computer Methods in Biomechanics and Biomedical Engineering, 1999, 2, 83-105.	1.6	16
103	Efficacy of routine fetal ultrasound screening for congenital heart disease in IsÃ¨re county, France. , 1999, 19, 318-322.		37
104	Characterization of a Germline Mosaicism in Families with Lowe Syndrome, and Identification of Seven Novel Mutations in the OCRL1 Gene. American Journal of Human Genetics, 1999, 65, 68-76.	6.2	52
105	Cytomegalovirus seroprevalence in French pregnant women: parity and place of birth as major predictive factors. European Journal of Epidemiology, 1998, 14, 147-152.	5.7	82
106	Population screening for aneuploidy using maternal age and ultrasound. , 1998, 18, 683-692.		13
107	Mapping of the orientation of myocardial cells by means of polarized light and confocal scanning laser microscopy. Microscopy Research and Technique, 1995, 30, 480-490.	2.2	40
108	Method for the study of the threeâ€dimensional orientation of the nuclei of myocardial cells in fetal human heart by means of confocal scanning laser microscopy. Journal of Microscopy, 1994, 174, 101-110.	1.8	24

#	Article	IF	Citations
109	Abnormal direct entry of the umbilical vein into the right atrium: antenatal detection, embryologic aspects. Surgical and Radiologic Anatomy, 1991, 13, 59-62.	1.2	25
110	Prediction of outcome by prenatal Doppler analysis in a patient with aortic stenosis.. Heart, 1991, 65, 53-54.	2.9	3
111	The crooked neck dwarf muscular dysgenesis in fowl is due to a selective alteration of the somitic myogenic cell line. Roux's Archives of Developmental Biology, 1988, 197, 49-55.	1.2	4