Simone Gana

List of Publications by Year
in descending order

19	424	1163117 8	839539 18
papers	citations	h-index	g-index

19	19	19	1169
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Heterozygous missense mutations in SMARCA2 cause Nicolaides-Baraitser syndrome. Nature Genetics, 2012, 44, 445-449.	21.4	207
2	19q13.11 cryptic deletion: description of two new cases and indication for a role of WTIP haploinsufficiency in hypospadias. European Journal of Human Genetics, 2012, 20, 852-856.	2.8	40
3	Genotypeâ€“phenotype correlates in Joubert syndrome: A review. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, 190, 72-88.	1.6	37
4	MEF2C deletions and mutations versus duplications: A clinical comparison. European Journal of Medical Genetics, 2013, 56, 260-265.	1.3	24
5	Loss-of-FunctionFANCLMutations Associate with Severe Fanconi Anemia Overlapping the VACTERL Association. Human Mutation, 2015, 36, 562-568.	2.5	23
6	SUFU haploinsufficiency causes a recognisable neurodevelopmental phenotype at the mild end of the Joubert syndrome spectrum. Journal of Medical Genetics, 2022, 59, 888-894.	3.2	19
7	Corpus callosum agenesis, severe mental retardation, epilepsy, and dyskinetic quadriparesis due to a novel mutation in the homeodomain of ARX. , 2011, 155, 892-897.		11
8	Congenital diaphragmatic hernia as prenatal presentation of Apert syndrome. Prenatal Diagnosis, 2011, 31, 910-911.	2.3	9
9	Nicolaidesâ€“Baraitser syndrome. Clinical Dysmorphology, 2011, 20, 38-41.	0.3	9
10	Familial Interstitial 6q23.2 Deletion Including Eya4 Associated With Otofaciocervical Syndrome. Frontiers in Genetics, 2019, 10, 650.	2.3	9
11	Blepharophimosis, Ptosis, Epicanthus Inversus Syndrome: New Report with a 197-kb Deletion Upstream of FOXL2 and Review of the Literature. Molecular Syndromology, 2019, 10, 147-153.	0.8	9
12	Sydenham's chorea in a girl with juvenile idiopathic arthritis treated with antiâ€TNFÎ± therapy. Movement Disorders, 2010, 25, 511-514.	3.9	5
13	Prenatal phenotype of Nager syndrome and Rodriguez syndrome. Clinical Dysmorphology, 2013, 22, 135-139.	0.3	5
14	<scp>PUS3</scp>â€related disorder: Report of a novel patient and delineation of the phenotypic spectrum. American Journal of Medical Genetics, Part A, 2022, 188, 635-641.	1.2	5
15	Novel insights into the clinico-radiological spectrum of phenotypes associated to PIGN mutations. European Journal of Paediatric Neurology, 2021, 33, 21-28.	1.6	4
16	Alazami syndrome: Phenotypic expansion and clinical resemblance to Smithâ€“Lemliâ€“Opitz syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2722-2726.	1.2	3
17	3p26.3 terminal deletions: a challenge for prenatal genetic counseling. Prenatal Diagnosis, 2017, 37, 197-200.	2.3	2
18	Advantages of Array Comparative Genomic Hybridization Using Buccal Swab DNA for Detecting Pallister-Killian Syndrome. Annals of Laboratory Medicine, 2019, 39, 232-234.	2.5	2

#	Article	IF	Citations
19	Extremely sustained startle-induced clonus: Non epileptic motor attacks mimicking clonic seizures in children with encephalopathy. Seizure: the Journal of the British Epilepsy Association, 2012, 21, 147-150.	2.0	1