# 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 | 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 |
| 2 | <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 |
| 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 | 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 |
| 5 | 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 |
| 6 | Familial Interstitial 6q23.2 Deletion Including Eya4 Associated With Otofaciocervical Syndrome. Frontiers in Genetics, 2019, 10, 650. | 2.3 | 9 |
| 7 | 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 |
| 8 | 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 |
| 9 | 3p26.3 terminal deletions: a challenge for prenatal genetic counseling. Prenatal Diagnosis, 2017, 37, 197-200. | 2.3 | 2 |
| 10 | Loss-of-FunctionFANCLMutations Associate with Severe Fanconi Anemia Overlapping the VACTERL Association. Human Mutation, 2015, 36, 562-568. | 2.5 | 23 |
| 11 | MEF2C deletions and mutations versus duplications: A clinical comparison. European Journal of Medical Genetics, 2013, 56, 260-265. | 1.3 | 24 |
| 12 | Prenatal phenotype of Nager syndrome and Rodriguez syndrome. Clinical Dysmorphology, 2013, 22, 135-139. | 0.3 | 5 |
| 13 | 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 |
| 14 | Heterozygous missense mutations in SMARCA2 cause Nicolaides-Baraitser syndrome. Nature Genetics, 2012, 44, 445-449. | 21.4 | 207 |
| 15 | 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 |
| 16 | Congenital diaphragmatic hernia as prenatal presentation of Apert syndrome. Prenatal Diagnosis, 2011, 31, 910-911. | 2.3 | 9 |
| 17 | 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 |
| 18 | Nicolaidesâ€“Baraitser syndrome. Clinical Dysmorphology, 2011, 20, 38-41. | 0.3 | 9 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Sydenham's chorea in a girl with juvenile idiopathic arthritis treated with antiâ€TNFÎ± therapy. Movement Disorders, 2010, 25, 511-514. | 3.9 | 5 |