Goran Cuturilo

List of Publications by Year
in descending order

28	588	687363 13	642732 23
papers	citations	h-index	g-index

28	28	28	1300
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Recessive DNAH9 Loss-of-Function Mutations Cause Laterality Defects and Subtle Respiratory Ciliary-Beating Defects. American Journal of Human Genetics, 2018, 103, 995-1008.	6.2	92
2	Phenotype and genotype of 87 patients with Mowatâ€“Wilson syndrome and recommendations for care. Genetics in Medicine, 2018, 20, 965-975.	2.4	67
3	PITX2 deficiency and associated human disease: insights from the zebrafish model. Human Molecular Genetics, 2018, 27, 1675-1695.	2.9	64
4	Comprehensive use of extended exome analysis improves diagnostic yield in rare disease: a retrospective survey in 1,059 cases. Genetics in Medicine, 2018, 20, 303-312.	2.4	57
5	Neuroimaging findings in Mowatâ€“Wilson syndrome: a study of 54 patients. Genetics in Medicine, 2017, 19, 691-700.	2.4	45
6	Activating Mutations of RRAS2 Are a Rare Cause of Noonan Syndrome. American Journal of Human Genetics, 2019, 104, 1223-1232.	6.2	43
7	Structural, Functional, and Clinical Characterization of a NovelPTPN11Mutation Cluster Underlying Noonan Syndrome. Human Mutation, 2017, 38, 451-459.	2.5	39
8	Rare missense TUBGCP5 gene variant in a patient with primary microcephaly. European Journal of Medical Genetics, 2019, 62, 103598.	1.3	22
9	Diagnostic and Clinical Utility of Clinical Exome Sequencing in Children With Moderate and Severe Global Developmental Delay / Intellectual Disability. Journal of Child Neurology, 2020, 35, 116-131.	1.4	22
10	4q34.1â€“q35.2 deletion in a boy with phenotype resembling 22q11.2 deletion syndrome. European Journal of Pediatrics, 2011, 170, 1465-1470.	2.7	19
11	The role of modern imaging techniques in the diagnosis of malposition of the branch pulmonary arteries and possible association with microdeletion 22q11.2. Cardiology in the Young, 2013, 23, 181-188.	0.8	19
12	Differences in speech and language abilities between children with 22q11.2 deletion syndrome and children with phenotypic features of 22q11.2 deletion syndrome but without microdeletion. Research in Developmental Disabilities, 2016, 55, 322-329.	2.2	17
13	The novel RAF1 mutation p.(Gly361Ala) located outside the kinase domain of the CR3 region in two patients with Noonan syndrome, including one with a rare brain tumor. American Journal of Medical Genetics, Part A, 2018, 176, 470-476.	1.2	17
14	Mowat-Wilson syndrome: growth charts. Orphanet Journal of Rare Diseases, 2020, 15, 151.	2.7	12
15	The Impact of 22q11.2 Microdeletion on Cardiac Surgery Postoperative Outcome. Pediatric Cardiology, 2017, 38, 1680-1685.	1.3	10
16	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. Genetics in Medicine, 2022, 24, 1283-1296.	2.4	9
17	A rare association of interrupted aortic arch type C and microdeletion 22q11.2. European Journal of Pediatrics, 2008, 167, 1195-1198.	2.7	7
18	The clinical significance of A2ML1 variants in Noonan syndrome has to be reconsidered. European Journal of Human Genetics, 2021, 29, 524-527.	2.8	7

#	Article	IF	Citations
19	Prenatal growth retardation, microcephaly, and eye coloboma in infant with multiple congenital anomalies: Further delineation of presumed new dysmorphic syndrome. Birth Defects Research Part A: Clinical and Molecular Teratology, 2008, 82, 166-168.	1.6	5
20	Improving the diagnosis of children with 22q11.2 deletion syndrome: A single-center experience from Serbia. Indian Pediatrics, 2016, 53, 786-789.	0.4	3
21	The landscape of Mucopolysaccharidosis in Southern and Eastern European countries: a survey from 19 specialistic centers. Orphanet Journal of Rare Diseases, 2022, 17, 136.	2.7	3
22	Ectodermal Defects and Anal Atresia in a Child with a <scp>TP</scp>63 Mutationâ€Expanding the Phenotypic Spectrum. Pediatric Dermatology, 2015, 32, 421-422.	0.9	2
23	Clientsâ€™ Perception of Outcome of Teamâ€Based Prenatal and Reproductive Genetic Counseling in Serbian Service Using the Perceived Personal Control (PPC) Questionnaire. Journal of Genetic Counseling, 2016, 25, 189-197.	1.6	2
24	Genetic evaluation of newborns with critical congenital heart defects admitted to the intensive care unit. European Journal of Pediatrics, 2021, 180, 3219-3227.	2.7	2
25	Rare Pathogenic Copy Number Variation in the 16p11.2 (BP4â€“BP5) Region Associated with Neurodevelopmental and Neuropsychiatric Disorders: A Review of the Literature. International Journal of Environmental Research and Public Health, 2020, 17, 9253.	2.6	1
26	Speech and language abilities of children with the familial form of 22q11.2 deletion syndrome. Genetika, 2016, 48, 57-72.	0.4	1
27	Ring chromosome 20: A further contribution to the delineation of epileptic phenotype. Vojnosanitetski Pregled, 2022, 79, 196-200.	0.2	1
28	Spina bifida occulta-a diagnostic feature of the trisomy 8 syndrome. Acta Chirurgica Iugoslavica, 2014, 61, 59-63.	0.0	0