Maria Gnazzo

List of Publications by Year
in descending order

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18 papers	400 citations	1040056 9 h-index	888059 17 g-index
18 all docs	18 docs citations	18 times ranked	926 citing authors

#	Article	IF	Citations
1	Clinical refinement of the <scp>SETD5</scp>â€associated phenotype in a child displaying novel features and <scp>KBG</scp> syndromeâ€like appearance. American Journal of Medical Genetics, Part A, 2022, 188, 1623-1625.	1.2	3
2	Congenital heart defects in molecularly confirmed <scp>KBG</scp> syndrome patients. American Journal of Medical Genetics, Part A, 2022, 188, 1149-1159.	1.2	5
3	Facial Dysmorphisms, Macrodontia, Focal Epilepsy, and Thinning of the Corpus Callosum: A Rare Mild Form of Kabuki Syndrome. Journal of Pediatric Genetics, 2021, 10, 049-052.	0.7	1
4	Clinical delineation, sex differences, and genotypeâ€“phenotype correlation in pathogenic KDM6A variants causing X-linked Kabuki syndrome type 2. Genetics in Medicine, 2021, 23, 1202-1210.	2.4	30
5	Cognitive and Adaptive Characterization of Children and Adolescents with KBG Syndrome: An Explorative Study. Journal of Clinical Medicine, 2021, 10, 1523.	2.4	2
6	Evolocumab in the management of children <10 years of age affected by homozygous familial hypercholesterolemia. Atherosclerosis, 2021, 324, 148-150.	0.8	2
7	A new missense mutation in DPF2 gene related to Coffin Siris syndrome 7: Description of a mild phenotype expanding DPF2-related clinical spectrum and differential diagnosis among similar syndromes epigenetically determined. Brain and Development, 2020, 42, 192-198.	1.1	3
8	KBG syndrome: Common and uncommon clinical features based on 31 new patients. American Journal of Medical Genetics, Part A, 2020, 182, 1073-1083.	1.2	27
9	Obsessive Compulsive Symptoms and Psychopathological Profile in Children and Adolescents with KBG Syndrome. Brain Sciences, 2019, 9, 313.	2.3	7
10	Novel Mutations and Unreported Clinical Features in KBG Syndrome. Molecular Syndromology, 2019, 10, 130-138.	0.8	23
11	Burkitt lymphoma in a patient with Kabuki syndrome carrying a novel KMT2D mutation. American Journal of Medical Genetics, Part A, 2019, 179, 113-117.	1.2	10
12	Treatment of homozygous familial hypercholesterolaemia in paediatric patients: A monocentric experience. European Journal of Preventive Cardiology, 2018, 25, 1098-1105.	1.8	5
13	Prominent and elongated coccyx, a new manifestation of KBG syndrome associated with novel mutation in ANKRD11. American Journal of Medical Genetics, Part A, 2018, 176, 1991-1995.	1.2	10
14	Congenital heart defects in molecularly proven Kabuki syndrome patients. American Journal of Medical Genetics, Part A, 2017, 173, 2912-2922.	1.2	60
15	Extensive Molecular Analysis Suggested the Strong Genetic Heterogeneity of Idiopathic Chronic Pancreatitis. Molecular Medicine, 2016, 22, 300-309.	4.4	17
16	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. Human Mutation, 2015, 36, 1080-1087.	2.5	67
17	Kabuki syndrome: clinical and molecular diagnosis in the first year of life. Archives of Disease in Childhood, 2015, 100, 158-164.	1.9	69
18	Diagnosis of Noonan syndrome and related disorders using target next generation sequencing. BMC Medical Genetics, 2014, 15, 14.	2.1	59