

Mattia Gentile

List of Publications by Year in descending order

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Version: 2024-02-01

25
papers

585
citations

687363

13
h-index

642732

23
g-index

25
all docs

25
docs citations

25
times ranked

600
citing authors

#	ARTICLE	IF	CITATIONS
1	12q21 Interstitial Deletions: Seven New Syndromic Cases Detected by Array-CGH and Review of the Literature. <i>Genes</i> , 2022, 13, 780.	2.4	0
2	Functional evidence of $mTOR1$ splice variant involvement in the pathogenesis of congenital heart defects. <i>Clinical Genetics</i> , 2021, 99, 425-429.	2.0	1
3	First prenatal case of Noonan syndrome with SOS2 mutation: Implications of early diagnosis for genetic counseling. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1897-1902.	1.2	1
4	Cytogenetic and Array-CGH Characterization of a Simple Case of Reciprocal t(3;10) Translocation Reveals a Hidden Deletion at 5q12. <i>Genes</i> , 2021, 12, 877.	2.4	1
5	Genetic Pattern, Orthodontic and Surgical Management of Multiple Supplementary Impacted Teeth in a Rare, Cleidocranial Dysplasia Patient: A Case Report. <i>Medicina (Lithuania)</i> , 2021, 57, 1350.	2.0	22
6	14q12q13.2 microdeletion syndrome: Clinical characterization of a new patient, review of the literature, and further evidence of a candidate region for CNS anomalies. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1289.	1.2	5
7	Novel exostosin-2 missense variants in a family with autosomal recessive exostosin-2-related syndrome: further evidences on the phenotype. <i>Clinical Genetics</i> , 2019, 95, 165-171.	2.0	3
8	Genomic changes of chromosomes 8p23.1 and 1q21: Novel mutations in malignant mesothelioma. <i>Lung Cancer</i> , 2018, 126, 106-111.	2.0	26
9	Peritoneal Mesothelioma with Residential Asbestos Exposure. Report of a Case with Long Survival (Seventeen Years) Analyzed by Cgh-Array. <i>International Journal of Molecular Sciences</i> , 2017, 18, 1818.	4.1	26
10	14q13 distal microdeletion encompassing <i>NKX2-1</i> and <i>PAX9</i> : Patient report and refinement of the associated phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1884-1888.	1.2	10
11	Molecular cytogenetic characterization and genotype/phenotype analysis in a patient with a de novo 8p23.2p23.3 deletion/12p13.31p13.33 duplication. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 1713-1718.	1.2	5
12	Non-syndromic multiple supernumerary teeth in a family unit with a normal karyotype: case report. <i>International Journal of Medical Sciences</i> , 2010, 7, 378-384.	2.5	64
13	Characterization of a complex chromosome aberration in two cases of peritoneal mesothelioma arising primarily in the hernial sac. <i>Pathology International</i> , 2009, 59, 415-421.	1.3	9
14	Holt-Oram syndrome associated with anomalies of the feet. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 1185-1189.	1.2	18
15	Prenatal diagnosis of chromosome 4 mosaicism: Prognostic role of cytogenetic, molecular, and ultrasound/MRI characterization. <i>American Journal of Medical Genetics, Part A</i> , 2005, 136A, 66-70.	1.2	15
16	FISH approach to determine cat eye syndrome chromosome breakpoints of a patient with cat eye syndrome type II. <i>European Journal of Medical Genetics</i> , 2005, 48, 33-39.	1.3	5
17	Cat-eye syndrome in a fetus with increased nuchal translucency: three-dimensional ultrasound and echocardiographic evaluation of the fetal phenotype. <i>Ultrasound in Obstetrics and Gynecology</i> , 2004, 24, 485-487.	1.7	16
18	Characteristics, associations and outcome of absent pulmonary valve syndrome in the fetus. <i>Ultrasound in Obstetrics and Gynecology</i> , 2004, 24, 623-628.	1.7	69

#	ARTICLE	IF	CITATIONS
19	Clinical, cytogenetic, and molecular characterization of a patient with a de novo interstitial 22q12 duplication. American Journal of Medical Genetics Part A, 2004, 127A, 186-190.	2.4	7
20	FISH and cytogenetic characterization of a terminal chromosome 1q deletion: Clinical case report and phenotypic implications. American Journal of Medical Genetics Part A, 2003, 117A, 251-254.	2.4	42
21	22q11 deletions in fetuses with malformations of the outflow tracts or interruption of the aortic arch: impact of additional ultrasound signs. Prenatal Diagnosis, 2003, 23, 752-757.	2.3	66
22	Prenatal diagnosis of interruption of the aortic arch and its association with deletion of chromosome 22q11. Ultrasound in Obstetrics and Gynecology, 2002, 20, 327-331.	1.7	34
23	Pulmonary alveolar microlithiasis: Clinical features, evolution of the phenotype, and review of the literature. American Journal of Medical Genetics Part A, 2002, 111, 220-224.	2.4	63
24	Interrupted aortic arch type A with 22q11 deletion: prenatal detection of an unusual association. Prenatal Diagnosis, 2002, 22, 371-374.	2.3	12
25	Prenatal diagnosis of ductus venosus agenesis and its association with cytogenetic/congenital anomalies. Prenatal Diagnosis, 2002, 22, 995-1000.	2.3	65