

# Jelena Martinovic

## List of Publications by Year in descending order

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Version: 2024-02-01

22  
papers

578  
citations

759233

12  
h-index

677142

22  
g-index

22  
all docs

22  
docs citations

22  
times ranked

1369  
citing authors

#	ARTICLE	IF	CITATIONS
1	Bi-allelic MYH3 loss-of-function variants cause a lethal form of contractures, pterygia, and spondylcarpotarsal fusion syndrome 1B. <i>Neuromuscular Disorders</i> , 2022, 32, 445-449.	0.6	2
2	Prenatal Diagnosis of <i>COL4A1</i> Mutations in Eight Cases: Further Delineation of the Neurohistopathological Phenotype. <i>Pediatric and Developmental Pathology</i> , 2022, 25, 435-446.	1.0	1
3	Discovering a new part of the phenotypic spectrum of Coffin-Siris syndrome in a fetal cohort. <i>Genetics in Medicine</i> , 2022, 24, 1753-1760.	2.4	6
4	The male external urethral sphincter is autonomically innervated. <i>Clinical Anatomy</i> , 2021, 34, 263-271.	2.7	11
5	Early prenatal diagnosis of alveolar capillary dysplasia with misalignment of pulmonary veins due to a 16q24.1 deletion. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1494-1497.	1.2	3
6	Dysregulation of the NRG1/ERBB pathway causes a developmental disorder with gastrointestinal dysmotility in humans. <i>Journal of Clinical Investigation</i> , 2021, 131, .	8.2	24
7	Human neuropathology confirms projection neuron and interneuron defects and delayed oligodendrocyte production and maturation in FOXP1 syndrome. <i>European Journal of Medical Genetics</i> , 2021, 64, 104282.	1.3	4
8	Two separate placental masses on ultrasound do not always indicate a dichorionic pregnancy. <i>Journal of Gynecology Obstetrics and Human Reproduction</i> , 2020, 49, 101694.	1.3	1
9	Severe and progressive neuronal loss in myelomeningocele begins before 16 weeks of pregnancy. <i>American Journal of Obstetrics and Gynecology</i> , 2020, 223, 256.e1-256.e9.	1.3	15
10	Complex Compound Inheritance of Lethal Lung Developmental Disorders Due to Disruption of the TBX-FGF Pathway. <i>American Journal of Human Genetics</i> , 2019, 104, 213-228.	6.2	90
11	Loss-of-function mutations in <i>KIF14</i> cause severe microcephaly and kidney development defects in humans and zebrafish. <i>Human Molecular Genetics</i> , 2019, 28, 778-795.	2.9	33
12	First prenatal case of proximal 19p13.12 microdeletion syndrome: New insights and new delineation of the syndrome. <i>European Journal of Medical Genetics</i> , 2018, 61, 322-328.	1.3	3
13	Detailed muscular structure and neural control anatomy of the levator ani muscle: a study based on female human fetuses. <i>American Journal of Obstetrics and Gynecology</i> , 2018, 218, 121.e1-121.e12.	1.3	22
14	Fetal cerebral hemorrhage due to X-linked <i>GATA1</i> gene mutation. <i>Prenatal Diagnosis</i> , 2018, 38, 772-778.	2.3	5
15	Congenital diaphragmatic hernia has a better prognosis when associated with a hernia sac. <i>Prenatal Diagnosis</i> , 2018, 38, 638-644.	2.3	15
16	Molecular diagnosis of PIK3CA-related overgrowth spectrum (PROS) in 162 patients and recommendations for genetic testing. <i>Genetics in Medicine</i> , 2017, 19, 989-997.	2.4	90
17	Targeted Exome Sequencing Identifies PBX1 as Involved in Monogenic Congenital Anomalies of the Kidney and Urinary Tract. <i>Journal of the American Society of Nephrology: JASN</i> , 2017, 28, 2901-2914.	6.1	90
18	Mutations in GREB1L Cause Bilateral Kidney Agenesis in Humans and Mice. <i>American Journal of Human Genetics</i> , 2017, 101, 803-814.	6.2	76

#	ARTICLE	IF	CITATIONS
19	WDR81 mutations cause extreme microcephaly and impair mitotic progression in human fibroblasts and Drosophila neural stem cells. <i>Brain</i> , 2017, 140, 2597-2609.	7.6	28
20	Levator ani muscle innervation: Anatomical study in human fetus. <i>Neurourology and Urodynamics</i> , 2017, 36, 1464-1471.	1.5	18
21	Increased TGF- $\beta$ : a drawback of tracheal occlusion in human and experimental congenital diaphragmatic hernia?. <i>American Journal of Physiology - Lung Cellular and Molecular Physiology</i> , 2016, 310, L311-L327.	2.9	15
22	Rare ACTG1 variants in fetal microlissencephaly. <i>European Journal of Medical Genetics</i> , 2015, 58, 416-418.	1.3	26