Gos Monika

List of Publications by Year
in descending order

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24 papers	1,081 citations	623734 14 h-index	477307 29 g-index
31 all docs	31 docs citations	31 times ranked	2806 citing authors

#	Article	IF	Citations
1	Floppy infant syndrome as a first manifestation of LMNA-related congenital muscular dystrophy. European Journal of Paediatric Neurology, 2021, 32, 115-121.	1.6	4
2	Bullous diseases caused by KRT1 gene mutations: from epidermolytic hyperkeratosis to a novel variant of epidermolysis bullosa simplex. Postepy Dermatologii I Alergologii, 2021, 38, 1032-1038.	0.9	2
3	The haploinsufficiency syndrome associated with de novo nonsense variant (P.GLN1981*). Medycyna Wieku Rozwojowego, 2021, 24, 32-36.	0.2	1
4	Successful Salvage Treosulfan-Based Megachemotherapy With Allogeneic Stem Cell Transplantation in Nonsyndromic, Therapy-Resistant Disseminated Juvenile Xanthogranuloma: A Case Report. Transplantation Proceedings, 2020, 52, 2844-2848.	0.6	4
5	The remarkable phenotypic variability of the p.Arg269HiS variant in the TRPV4 gene. Muscle and Nerve, 2019, 59, 129-133.	2.2	8
6	Splicing mutations in human genetic disorders: examples, detection, and confirmation. Journal of Applied Genetics, 2018, 59, 253-268.	1.9	426
7	Craniosynostosis as a clinical and diagnostic problem: molecular pathology and genetic counseling. Journal of Applied Genetics, 2018, 59, 133-147.	1.9	45
8	MAP2K2 mutation as a cause of cardioâ€facioâ€cutaneous syndrome in an infant with a severe and fatal course of the disease. American Journal of Medical Genetics, Part A, 2018, 176, 1670-1674.	1.2	7
9	Novel Col12A1 variant expands the clinical picture of congenital myopathies with extracellular matrix defects. Muscle and Nerve, 2017, 55, 277-281.	2.2	31
10	Towards a Better Molecular Diagnosis of FMR1-Related Disordersâ€"A Multiyear Experience from a Reference Lab. Genes, 2016, 7, 59.	2.4	4
11	Monoallelic and biallelic deletions of 13q14 in a group of CLL/SLL patients investigated by CGH Haematological Cancer and SNP array (8x60K). Molecular Cytogenetics, 2016, 9, 1.	0.9	32
12	Atypical fibrodysplasia ossificans progressiva diagnosed by wholeâ€exome sequencing. American Journal of Medical Genetics, Part A, 2015, 167, 1337-1341.	1.2	11
13	Rare variants inSOS2andLZTR1are associated with Noonan syndrome. Journal of Medical Genetics, 2015, 52, 413-421.	3.2	187
14	Rett-like onset in late-infantile neuronal ceroid lipofuscinosis (CLN7) caused by compound heterozygous mutation in the MFSD8 gene and review of the literature data on clinical onset signs. European Journal of Paediatric Neurology, 2015, 19, 78-86.	1.6	25
15	Novel point mutations in survival motor neuron 1 gene expand the spectrum of phenotypes observed in spinal muscular atrophy patients. Neuromuscular Disorders, 2014, 24, 617-623.	0.6	27
16	Contribution of RIT1 mutations to the pathogenesis of Noonan syndrome: Four new cases and further evidence of heterogeneity. American Journal of Medical Genetics, Part A, 2014, 164, 2310-2316.	1.2	42
17	Severe phenotypes of SMARD1 associated with novel mutations of the IGHMBP2 gene and nuclear degeneration of muscle and Schwann cells. European Journal of Paediatric Neurology, 2014, 18, 183-192.	1.6	20
18	Epigenetic mechanisms of gene expression regulation in neurological diseases. Acta Neurobiologiae Experimentalis, 2013, 73, 19-37.	0.7	21

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19	Application of array comparative genomic hybridization in 102 patients with epilepsy and additional neurodevelopmental disorders. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2012, 159B, 760-771.	1.7	48
20	Early-onset seizures due to mosaic exonic deletions of CDKL5 in a male and two females. Genetics in Medicine, 2011, 13, 447-452.	2.4	45
21	Molecular signature of cell cycle exit induced in human T lymphoblasts by IL-2 withdrawal. BMC Genomics, 2009, 10, 261.	2.8	17
22	Identification of mutations in theNF2 gene in Polish patients with neurofibromatosis type 2. Journal of Applied Genetics, 2008, 49, 297-300.	1.9	3
23	Unusual Cyclin D1 Positive Marginal Zone Lymphoma of Mediastinum. Medical Oncology, 2006, 23, 423-428.	2.5	8
24	Mantle Cell Lymphoma Presenting with Paraproteinemia. Medical Oncology, 2005, 22, 319-324.	2.5	6