Anna Elsa Maria Allegri

List of Publications by Year
in descending order

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Version: 2024-02-01

32	933	516561 16	454834 30
papers	citations	h-index	g-index

33	33	33	1231
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Osteogenesis Imperfecta/Ehlersâ€“Danlos Overlap Syndrome and Neuroblastomaâ€”Case Report and Review of Literature. Genes, 2022, 13, 581.	1.0	2
2	Maternal Uniparental Disomy of Chromosome 20 (UPD(20)mat) as Differential Diagnosis of Silver Russell Syndrome: Identification of Three New Cases. Genes, 2021, 12, 588.	1.0	6
3	Cognitive and White Matter Microstructure Development in Congenital Hypothyroidism and Familial Thyroid Disorders. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e3990-e4006.	1.8	10
4	Novel compound heterozygous pathogenic variants in nucleotide-binding protein like protein (NUBPL) cause leukoencephalopathy with multi-systemic involvement. Molecular Genetics and Metabolism, 2020, 129, 26-34.	0.5	9
5	Endocrine Outcomes In Central Diabetes Insipidus: the Predictive Value of Neuroimaging â€œMismatch Patternâ€. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 3562-3574.	1.8	3
6	Antibodies Against Hypothalamus and Pituitary Gland in Childhood-Onset Brain Tumors and Pituitary Dysfunction. Frontiers in Endocrinology, 2020, 11, 16.	1.5	9
7	Accuracy and Limitations of the Growth Hormone (GH) Releasing Hormone-Arginine Retesting in Young Adults With Childhood-Onset GH Deficiency. Frontiers in Endocrinology, 2019, 10, 525.	1.5	10
8	Familial ROBO1 deletion associated with ectopic posterior pituitary, duplication of the pituitary stalk and anterior pituitary hypoplasia. Journal of Pediatric Endocrinology and Metabolism, 2019, 32, 95-99.	0.4	11
9	Central adrenal insufficiency in children and adolescents. Best Practice and Research in Clinical Endocrinology and Metabolism, 2018, 32, 425-444.	2.2	45
10	Role of MRI T2-DRIVE in the assessment of pituitary stalk abnormalities without gadolinium in pituitary diseases. European Journal of Endocrinology, 2018, 178, 613-622.	1.9	22
11	A novel pathogenic MYH3 mutation in a child with Sheldonâ€“Hall syndrome and vertebral fusions. American Journal of Medical Genetics, Part A, 2018, 176, 663-667.	0.7	16
12	Novel CNS malformations and skeletal anomalies in a patient with Beaulieuâ€boycottâ€innes syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2835-2840.	0.7	9
13	Reliability of clonidine testing for the diagnosis of growth hormone deficiency in children and adolescents. Clinical Endocrinology, 2018, 89, 765-770.	1.2	6
14	Early impairment of somatosensory evoked potentials in very young children with achondroplasia with foramen magnum stenosis. Developmental Medicine and Child Neurology, 2017, 59, 192-198.	1.1	11
15	Classical and non-classical causes of GH deficiency in the paediatric age. Best Practice and Research in Clinical Endocrinology and Metabolism, 2016, 30, 705-736.	2.2	43
16	Age- and sex-matched reference curves for serum collagen type I C-telopeptides and bone alkaline phosphatase in children and adolescents: An alternative multivariate statistical analysis approach. Clinical Biochemistry, 2016, 49, 802-807.	0.8	16
17	Management of diabetes insipidus and adipsia in the child. Best Practice and Research in Clinical Endocrinology and Metabolism, 2015, 29, 415-436.	2.2	39
18	Association of achondroplasia with sagittal synostosis and scaphocephaly in two patients, an underestimated condition?. American Journal of Medical Genetics, Part A, 2015, 167, 646-652.	0.7	15

#	Article	IF	Citations
19	Early-onset central diabetes insipidus is associated with de novo arginine vasopressinâ€“neurophysin II or Wolfram syndrome 1 gene mutations. European Journal of Endocrinology, 2015, 172, 461-472.	1.9	24
20	Central Diabetes Insipidus in Children and Young Adults: Etiological Diagnosis and Long-Term Outcome of Idiopathic Cases. Journal of Clinical Endocrinology and Metabolism, 2014, 99, 1264-1272.	1.8	97
21	Midbrain-Hindbrain Involvement in Septo-Optic Dysplasia. American Journal of Neuroradiology, 2014, 35, 1586-1592.	1.2	22
22	Diabetes Insipidus â€“ Diagnosis and Management. Hormone Research in Paediatrics, 2012, 77, 69-84.	0.8	222
23	The use of neuroimaging for assessing disorders of pituitary development. Clinical Endocrinology, 2012, 76, 161-176.	1.2	62
24	Posterior pituitary (PP) evaluation in patients with anterior pituitary defect associated with ectopic PP and septo-optic dysplasia. European Journal of Endocrinology, 2011, 165, 411-420.	1.9	24
25	The Accuracy of the Glucagon Test Compared to the Insulin Tolerance Test in the Diagnosis of Adrenal Insufficiency in Young Children with Growth Hormone Deficiency. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 2132-2139.	1.8	38
26	Hepatocellular adenoma and metabolic balance in patients with type Ia glycogen storage disease. Molecular Genetics and Metabolism, 2008, 93, 398-402.	0.5	47
27	A case of ethylmalonic encephalopathy with atypical clinical and biochemical presentation. Molecular Genetics and Metabolism, 2006, 89, 395-397.	0.5	22
28	Multiple cryptic splice sites can be activated by IDS point mutations generating misspliced transcripts. Journal of Molecular Medicine, 2006, 84, 692-700.	1.7	21
29	Homozygosity for a non-pseudogene complex glucocerebrosidase allele as cause of an atypical neuronopathic form of Gaucher disease. American Journal of Medical Genetics, Part A, 2005, 134A, 95-96.	0.7	10
30	Congenital disorder of glycosylation (CDG) Ig: Report on a patient and review of the literature. Journal of Inherited Metabolic Disease, 2005, 28, 1162-1164.	1.7	18
31	Different Molecular Mechanisms Leading to White Matter Hypomyelination in Infantile Onset Lysosomal Disorders. Neuropediatrics, 2005, 36, 265-269.	0.3	34
32	MRI in acute intermittent maple syrup urine disease. Neurology, 2004, 63, 1078-1078.	1.5	8