

# Anna Elsa Maria Allegri

## List of Publications by Year in descending order

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32  
papers

933  
citations

516710

16  
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454955

30  
g-index

33  
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33  
docs citations

33  
times ranked

1231  
citing authors

#	ARTICLE	IF	CITATIONS
1	Diabetes Insipidus – Diagnosis and Management. <i>Hormone Research in Paediatrics</i> , 2012, 77, 69-84.	1.8	222
2	Central Diabetes Insipidus in Children and Young Adults: Etiological Diagnosis and Long-Term Outcome of Idiopathic Cases. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2014, 99, 1264-1272.	3.6	97
3	The use of neuroimaging for assessing disorders of pituitary development. <i>Clinical Endocrinology</i> , 2012, 76, 161-176.	2.4	62
4	Hepatocellular adenoma and metabolic balance in patients with type Ia glycogen storage disease. <i>Molecular Genetics and Metabolism</i> , 2008, 93, 398-402.	1.1	47
5	Central adrenal insufficiency in children and adolescents. <i>Best Practice and Research in Clinical Endocrinology and Metabolism</i> , 2018, 32, 425-444.	4.7	45
6	Classical and non-classical causes of GH deficiency in the paediatric age. <i>Best Practice and Research in Clinical Endocrinology and Metabolism</i> , 2016, 30, 705-736.	4.7	43
7	Management of diabetes insipidus and adipsia in the child. <i>Best Practice and Research in Clinical Endocrinology and Metabolism</i> , 2015, 29, 415-436.	4.7	39
8	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. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2010, 95, 2132-2139.	3.6	38
9	Different Molecular Mechanisms Leading to White Matter Hypomyelination in Infantile Onset Lysosomal Disorders. <i>Neuropediatrics</i> , 2005, 36, 265-269.	0.6	34
10	Posterior pituitary (PP) evaluation in patients with anterior pituitary defect associated with ectopic PP and septo-optic dysplasia. <i>European Journal of Endocrinology</i> , 2011, 165, 411-420.	3.7	24
11	Early-onset central diabetes insipidus is associated with de novo arginine vasopressin – neurophysin II or Wolfram syndrome 1 gene mutations. <i>European Journal of Endocrinology</i> , 2015, 172, 461-472.	3.7	24
12	A case of ethylmalonic encephalopathy with atypical clinical and biochemical presentation. <i>Molecular Genetics and Metabolism</i> , 2006, 89, 395-397.	1.1	22
13	Midbrain-Hindbrain Involvement in Septo-Optic Dysplasia. <i>American Journal of Neuroradiology</i> , 2014, 35, 1586-1592.	2.4	22
14	Role of MRI T2-DRIVE in the assessment of pituitary stalk abnormalities without gadolinium in pituitary diseases. <i>European Journal of Endocrinology</i> , 2018, 178, 613-622.	3.7	22
15	Multiple cryptic splice sites can be activated by IDS point mutations generating misspliced transcripts. <i>Journal of Molecular Medicine</i> , 2006, 84, 692-700.	3.9	21
16	Congenital disorder of glycosylation (CDG) Ig: Report on a patient and review of the literature. <i>Journal of Inherited Metabolic Disease</i> , 2005, 28, 1162-1164.	3.6	18
17	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. <i>Clinical Biochemistry</i> , 2016, 49, 802-807.	1.9	16
18	A novel pathogenic <i>MYH3</i> mutation in a child with Sheldon – Hall syndrome and vertebral fusions. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 663-667.	1.2	16

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19	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.	1.2	15
20	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.	2.1	11
21	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.	0.9	11
22	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.	1.2	10
23	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.	3.5	10
24	Cognitive and White Matter Microstructure Development in Congenital Hypothyroidism and Familial Thyroid Disorders. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e3990-e4006.	3.6	10
25	Novel CNS malformations and skeletal anomalies in a patient with Beaulieu-Boycott-Innes syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2835-2840.	1.2	9
26	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.	1.1	9
27	Antibodies Against Hypothalamus and Pituitary Gland in Childhood-Onset Brain Tumors and Pituitary Dysfunction. Frontiers in Endocrinology, 2020, 11, 16.	3.5	9
28	MRI in acute intermittent maple syrup urine disease. Neurology, 2004, 63, 1078-1078.	1.1	8
29	Reliability of clonidine testing for the diagnosis of growth hormone deficiency in children and adolescents. Clinical Endocrinology, 2018, 89, 765-770.	2.4	6
30	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.	2.4	6
31	Endocrine Outcomes In Central Diabetes Insipidus: the Predictive Value of Neuroimaging "Mismatch Pattern". Journal of Clinical Endocrinology and Metabolism, 2020, 105, 3562-3574.	3.6	3
32	Osteogenesis Imperfecta/Ehlers-Danlos Overlap Syndrome and Neuroblastoma Case Report and Review of Literature. Genes, 2022, 13, 581.	2.4	2