

Laura Audi

List of Publications by Year in descending order

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papers

916
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516710

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all docs

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times ranked

1007
citing authors

#	ARTICLE	IF	CITATIONS
1	Response to Letter to the Editor: "A Genome-Wide Pharmacogenetic Study of Growth Hormone Responsiveness". Journal of Clinical Endocrinology and Metabolism, 2021, 106, e409-e410.	3.6	0
2	A Genome-Wide Pharmacogenetic Study of Growth Hormone Responsiveness. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 3203-3214.	3.6	16
3	Molecular Basis of CYP19A1 Deficiency in a 46,XX Patient With R550W Mutation in <i>POR</i> : Expanding the PORD Phenotype. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e1272-e1290.	3.6	12
4	Reduced Androgen Receptor Expression in Genital Skin Fibroblasts From Patients With 45,X/46,XY Mosaicism. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 4630-4638.	3.6	5
5	GATA4 Variants in Individuals With a 46,XY Disorder of Sex Development (DSD) May or May Not Be Associated With Cardiac Defects Depending on Second Hits in Other DSD Genes. Frontiers in Endocrinology, 2018, 9, 142.	3.5	26
6	Nutritional rickets: vitamin D, calcium, and the genetic make-up. Pediatric Research, 2017, 81, 356-363.	2.3	6
7	Human MAMLD1 Gene Variations Seem Not Sufficient to Explain a 46,XY DSD Phenotype. PLoS ONE, 2015, 10, e0142831.	2.5	19
8	Past Experiences of Adults with Disorders of Sex Development. Endocrine Development, 2014, 27, 138-148.	1.3	5
9	<i>STAR</i> splicing mutations cause the severe phenotype of lipoid congenital adrenal hyperplasia: insights from a novel splice mutation and review of reported cases. Clinical Endocrinology, 2014, 80, 191-199.	2.4	15
10	Growth Hormone Receptor Polymorphism and Growth Hormone Therapy Response in Children: A Bayesian Meta-Analysis. American Journal of Epidemiology, 2012, 175, 867-877.	3.4	33
11	Contribution of human growth hormone-releasing hormone receptor (<i>GHRHR</i>) gene sequence variation to isolated severe growth hormone deficiency (<i>ISGHD</i>) and normal adult height. Clinical Endocrinology, 2012, 77, 564-574.	2.4	7
12	Pubertal Growth and Adult Height According to Age at Pubertal Growth Spurt Onset: Data from a Spanish Study Including 540 Subjects (281 Boys and 259 Girls). , 2012, , 1525-1544.		7
13	Pharmacogenomics of GH-Induced Catch-Up Growth in Prepubertal Short Children. The Role of the Exon 3-Deleted/Full-Length Growth Hormone Receptor Polymorphism. , 2012, , 917-933.		0
14	Methods to Study Cartilage and Bone Development. Endocrine Development, 2011, 21, 52-66.	1.3	12
15	Characterization of Novel StAR (Steroidogenic Acute Regulatory Protein) Mutations Causing Non-Classic Lipoid Adrenal Hyperplasia. PLoS ONE, 2011, 6, e20178.	2.5	75
16	Absence of GH-Releasing Hormone (GHRH) Mutations in Selected Patients with Isolated GH Deficiency. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E1457-E1460.	3.6	17
17	Loss of the C Terminus of Melanocortin Receptor 2 (MC2R) Results in Impaired Cell Surface Expression and ACTH Insensitivity. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E65-E72.	3.6	10
18	Vascular Endothelial Growth Factor Is Expressed in Human Fetal Growth Cartilage. Journal of Bone and Mineral Research, 2010, 15, 534-540.	2.8	56

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19	Longitudinal Pubertal Growth According to Age at Pubertal Growth Spurt Onset: Data from a Spanish Study Including 458 Children (223 Boys and 235 Girls). <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2009, 22, 715-26.	0.9	31
20	ACTH-dependent precocious pseudopuberty in an infant with DAX1 gene mutation. <i>European Journal of Pediatrics</i> , 2009, 168, 65-69.	2.7	12
21	Rickets in the Middle East: Role of Environment and Genetic Predisposition. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2008, 93, 1743-1750.	3.6	107
22	Growth Hormone (GH) Dose, But Not Exon 3-Deleted/Full-Length GH Receptor Polymorphism Genotypes, Influences Growth Response to Two-Year GH Therapy in Short Small-for-Gestational-Age Children. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2008, 93, 147-153.	3.6	44
23	Human growth hormone (<i>GH1</i>) gene polymorphism map in a normalâ€statured adult population. <i>Clinical Endocrinology</i> , 2007, 66, 258-268.	2.4	23
24	Exon 3-Deleted/Full-Length Growth Hormone Receptor Polymorphism Genotype Frequencies in Spanish Short Small-for-Gestational-Age (SGA) Children and Adolescents (n = 247) and in an Adult Control Population (n = 289) Show Increased <i>fl/fl</i> in Short SGA. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2006, 91, 5038-5043.	3.6	62
25	Genetically Determined Gonadal Tumours in Children. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2005, 18, 1215-1226.	0.9	0
26	SRD5A2 gene Q126R exon 2 point mutation in unrelated Spanish male pseudohermaphrodite patients.. <i>International Journal on Disability and Human Development</i> , 2005, 4, .	0.2	0
27	Clinical, Biochemical and Morphologic Diagnostic Markers in an Infant Male Pseudohermaphrodite Patient with Compound Heterozygous Mutations (G115D/R246W) in SRD5A2 Gene. <i>Hormone Research in Paediatrics</i> , 2004, 62, 259-264.	1.8	16
28	Areal Bone Mineral Density of the Lumbar Spine in 80 Premature Newborns. A Prospective and Longitudinal Study. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2004, 17, 959-66.	0.9	8
29	Compound Heterozygous Mutations in the <i>SRD5A2</i> Gene Exon 4 in a Male Pseudohermaphrodite Patient of Chinese Origin. <i>Journal of Andrology</i> , 2004, 25, 412-416.	2.0	38
30	Clinical and Biochemical Determinants of Bone Metabolism and Bone Mass in Adolescent Female Patients with Anorexia Nervosa. <i>Pediatric Research</i> , 2002, 51, 497-504.	2.3	111
31	Growth hormone/insulin-like growth factors axis in children undergoing cardiac surgery. <i>Critical Care Medicine</i> , 2001, 29, 1234-1238.	0.9	28
32	Anomalous Costochondral Cartilage in Fetal Anencephaly. <i>Pediatric and Developmental Pathology</i> , 2000, 3, 256-263.	1.0	1
33	Genetic Determinants of Bone Mass. <i>Hormone Research in Paediatrics</i> , 1999, 51, 105-123.	1.8	50
34	Down's Syndrome: Altered Chondrogenesis in Fetal Rib. <i>Pediatric Research</i> , 1998, 44, 93-98.	2.3	13
35	Morphologic and Metabolic Development of Human Fetal Epiphyseal Chondrocytes in Primary Culture. <i>Pediatric Research</i> , 1985, 19, 720-727.	2.3	13