Alexander A L Jorge

List of Publications by Year in Descending Order

155 papers	3,385 citations	34 h-index	51 g-index
172 ext. papers	4,149 ext. citations	4.1 avg, IF	5.02 L-index

#	Paper	IF	Citations
155	Cushing disease due to a somatic mutation in a patient with evolving pituitary hormone deficiencies due to a germline splicing variant.. *Archives of Endocrinology and Metabolism*, **2022**,	2.2	0
154	Idiopathic Short Stature: Diagnostic and Therapeutic Approach **2022**, 163-172		
153	Clinical and genetic characterization and long-term evaluation of individuals with maturity-onset diabetes of the young (MODY): the journey towards appropriate treatment.. *Diabetes Research and Clinical Practice*, **2022**, 109875	7.4	0
152	Targeted massively parallel sequencing for congenital generalized lipodystrophy. *Archives of Endocrinology and Metabolism*, **2021**, 64, 559-566	2.2	
151	Congenital chromoanagenesis in the routine postnatal chromosomal microarray analyses. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2335-2344	2.5	0
150	Adult Height in 299 Patients with Turner Syndrome with or without Growth Hormone Therapy: Results and Literature Review. *Hormone Research in Paediatrics*, **2021**, 94, 63-70	3.3	0
149	Genetics, clinical features and outcomes of non-syndromic pituitary gigantism: experience of a single center from Sao Paulo, Brazil. *Pituitary*, **2021**, 24, 252-261	4.3	
148	Activation of the MAPK pathway (RASopathies) and partial growth hormone insensitivity. *Molecular and Cellular Endocrinology*, **2021**, 519, 111040	4.4	8
147	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	5.6	
146	Insights from the genetic characterization of central precocious puberty associated with multiple anomalies. *Human Reproduction*, **2021**, 36, 506-518	5.7	6
145	SCUBE3 loss-of-function causes a recognizable recessive developmental disorder due to defective bone morphogenetic protein signaling. *American Journal of Human Genetics*, **2021**, 108, 115-133	11	8
144	Performance of mutation pathogenicity prediction tools on missense variants associated with 46,XY differences of sex development. *Clinics*, **2021**, 76, e2052	2.3	2
143	Disorders of Childhood Growth **2021**, 299-356		1
142	The phenotypic spectrum associated with OTX2 mutations in humans. *European Journal of Endocrinology*, **2021**, 185, 121-135	6.5	3
141	High-throughput splicing assays identify missense and silent splice-disruptive POU1F1 variants underlying pituitary hormone deficiency. *American Journal of Human Genetics*, **2021**, 108, 1526-1539	11	5
140	Variants in 46,XY DSD-Related Genes in Syndromic and Non-Syndromic Small for Gestational Age Children with Hypospadias. *Sexual Development*, **2021**, 1-7	1.6	0
139	Hormone resistance and short stature: A journey through the pathways of hormone signaling. *Molecular and Cellular Endocrinology*, **2021**, 536, 111416	4.4	0

138	Noonan syndrome patients beyond the obvious phenotype: A potential unfavorable metabolic profile. *American Journal of Medical Genetics, Part A*, **2021**, 185, 774-780	2.5	2
137	Growth Hormone insensitivity (Laron syndrome): Report of a new family and review of Brazilian patients. *Genetics and Molecular Biology*, **2020**, 42, e20180197	2	4
136	SELAdb: A database of exonic variants in a Brazilian population referred to a quaternary medical center in São Paulo. *Clinics*, **2020**, 75, e1913	2.3	3
135	Genetic investigation of patients with tall stature. *European Journal of Endocrinology*, **2020**, 182, 139-147	6.5	1
134	Clinical and Genetic Characterization of a Constitutional Delay of Growth and Puberty Cohort. *Neuroendocrinology*, **2020**, 110, 959-966	5.6	6
133	A Genome-Wide Pharmacogenetic Study of Growth Hormone Responsiveness. *Journal of Clinical Endocrinology and Metabolism*, **2020**, 105,	5.6	11
132	Phenotype-genotype analysis of 242 individuals with RASopathies: 18-year experience of a tertiary center in Brazil. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2020**, 184, 896-911	3.1	2
131	A Bayesian Approach to Diagnose Growth Hormone Deficiency in Children: Insulin-Like Growth Factor Type 1 Is Valuable for Screening and IGF-Binding Protein Type 3 for Confirmation. *Hormone Research in Paediatrics*, **2020**, 93, 197-205	3.3	3
130	Clinical and Molecular Description of 16 Families With Heterozygous IHH Variants. *Journal of Clinical Endocrinology and Metabolism*, **2020**, 105,	5.6	3
129	Diagnosis, Genetics, and Therapy of Short Stature in Children: A Growth Hormone Research Society International Perspective. *Hormone Research in Paediatrics*, **2019**, 92, 1-14	3.3	94
128	IGF-1 assessed by pubertal status has the best positive predictive power for GH deficiency diagnosis in peripubertal children. *Journal of Pediatric Endocrinology and Metabolism*, **2019**, 32, 173-179	1.6	16
127	Impact of Growth Hormone Therapy on Adult Height in Patients with PTPN11 Mutations Related to Noonan Syndrome. *Hormone Research in Paediatrics*, **2019**, 91, 252-261	3.3	6
126	Evaluation of SHOX defects in the era of next-generation sequencing. *Clinical Genetics*, **2019**, 96, 261-265	4	5
125	Exome Sequencing Reveals the POLR3H Gene as a Novel Cause of Primary Ovarian Insufficiency. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 2827-2841	5.6	17
124	Growth Failure and Experience With Growth Hormone Therapy in Noonan Syndrome **2019**, 15-29		
123	DLK1 Is a Novel Link Between Reproduction and Metabolism. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 2112-2120	5.6	44
122	Genetic causes of isolated short stature. *Archives of Endocrinology and Metabolism*, **2019**, 63, 70-78	2.2	17
121	Noonan syndrome associated with growth hormone deficiency with biallelic LZTR1 variants. *Genetics in Medicine*, **2019**, 21, 260	8.1	8

120	Two rare loss-of-function variants in the STAG3 gene leading to primary ovarian insufficiency. *European Journal of Medical Genetics*, **2019**, 62, 186-189	2.6	23
119	Combined pituitary hormone deficiency caused by PROP1 mutations: update 20 years post-discovery. *Archives of Endocrinology and Metabolism*, **2019**, 63, 167-174	2.2	12
118	Response to Letter to the Editor: "IHH Gene Mutations Causing Short Stature With Nonspecific Skeletal Abnormalities and Response to Growth Hormone Therapy". *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 5118-5119	5.6	1
117	Targeted sequencing identifies novel variants in common and rare MODY genes. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e962	2.3	12
116	Genetic Evidence of the Association of DEAH-Box Helicase 37 Defects With 46,XY Gonadal Dysgenesis Spectrum. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 5923-5934	5.6	14
115	Genetic Disorders in Prenatal Onset Syndromic Short Stature Identified by Exome Sequencing. *Journal of Pediatrics*, **2019**, 215, 192-198	3.6	18
114	Genotype and phenotype landscape of MEN2 in 554 medullary thyroid cancer patients: the BrasMEN study. *Endocrine Connections*, **2019**, 8, 289-298	3.5	14
113	Genetic diagnosis of congenital hypopituitarism by a target gene panel: novel pathogenic variants in GLI2, OTX2 and GHRHR. *Endocrine Connections*, **2019**, 8, 590-595	3.5	6
112	ESR1 polymorphism (rs2234693) influences femoral bone mass in patients with Turner syndrome. *Endocrine Connections*, **2019**, 8, 1513-1519	3.5	3
111	New genetic findings in a large cohort of congenital hypogonadotropic hypogonadism. *European Journal of Endocrinology*, **2019**, 181, 103-119	6.5	41
110	Update on new GH-IGF axis genetic defects. *Archives of Endocrinology and Metabolism*, **2019**, 63, 608-617	2.2	5
109	Multigene Sequencing Analysis of Children Born Small for Gestational Age With Isolated Short Stature. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 2023-2030	5.6	30
108	Growth and Clinical Characteristics of Children with Floating-Harbor Syndrome: Analysis of Current Original Data and a Review of the Literature. *Hormone Research in Paediatrics*, **2019**, 92, 115-123	3.3	4
107	Growth hormone therapy in children; research and practice - A review. *Growth Hormone and IGF Research*, **2019**, 44, 20-32	2	29
106	Oncogenic mutations in KEAP1 disturbing inhibitory Nrf2-Keap1 interaction: Activation of antioxidative pathway in papillary thyroid carcinoma. *Head and Neck*, **2018**, 40, 1271-1278	4.2	10
105	IHH Gene Mutations Causing Short Stature With Nonspecific Skeletal Abnormalities and Response to Growth Hormone Therapy. *Journal of Clinical Endocrinology and Metabolism*, **2018**, 103, 604-614	5.6	36
104	Pathogenic copy number variants in patients with congenital hypopituitarism associated with complex phenotypes. *Clinical Endocrinology*, **2018**, 88, 425-431	3.4	7
103	Genetic short stature. *Growth Hormone and IGF Research*, **2018**, 38, 29-33	2	22

102	PDX1 -MODY and dorsal pancreatic agenesis: New phenotype of a rare disease. *Clinical Genetics*, **2018**, 93, 382-386	4	15
101	Mutations in C-natriuretic peptide (NPPC): a novel cause of autosomal dominant short stature. *Genetics in Medicine*, **2018**, 20, 91-97	8.1	35
100	BCL11B mutations in patients affected by a neurodevelopmental disorder with reduced type 2 innate lymphoid cells. *Brain*, **2018**, 141, 2299-2311	11.2	36
99	Novel SUZ12 mutations in Weaver-like syndrome. *Clinical Genetics*, **2018**, 94, 461-466	4	22
98	Germline mutation landscape of multiple endocrine neoplasia type 1 using full gene next-generation sequencing. *European Journal of Endocrinology*, **2018**, 179, 391-407	6.5	6
97	Homozygous loss of function BRCA1 variant causing a Fanconi-anemia-like phenotype, a clinical report and review of previous patients. *European Journal of Medical Genetics*, **2018**, 61, 130-133	2.6	22
96	Recurrent Copy Number Variants Associated with Syndromic Short Stature of Unknown Cause. *Hormone Research in Paediatrics*, **2018**, 89, 13-21	3.3	17
95	Comparison of 68Ga PET/CT to Other Imaging Studies in Medullary Thyroid Cancer: Superiority in Detecting Bone Metastases. *Journal of Clinical Endocrinology and Metabolism*, **2018**, 103, 3250-3259	5.6	24
94	Homozygous and Heterozygous Nuclear Lamin A p.R582C Mutation: Different Lipodystrophic Phenotypes in the Same Kindred. *Frontiers in Endocrinology*, **2018**, 9, 458	5.7	8
93	Central Precocious Puberty Caused by a Heterozygous Deletion in the MKRN3 Promoter Region. *Neuroendocrinology*, **2018**, 107, 127-132	5.6	12
92	The recurrent PPP1CB mutation p.Pro49Arg in an additional Noonan-like syndrome individual: Broadening the clinical phenotype. *American Journal of Medical Genetics, Part A*, **2017**, 173, 824-828	2.5	18
91	Clinical application of ACMG-AMP guidelines in HNF1A and GCK variants in a cohort of MODY families. *Clinical Genetics*, **2017**, 92, 388-396	4	12
90	MANAGEMENT OF ENDOCRINE DISEASE: Diagnostic and therapeutic approach of tall stature. *European Journal of Endocrinology*, **2017**, 176, R339-R353	6.5	20
89	SOCS2 polymorphisms are not associated with clinical and biochemical phenotypes in acromegalic patients. *Pituitary*, **2017**, 20, 319-324	4.3	1
88	Long-term response to growth hormone therapy in a patient with short stature caused by a novel heterozygous mutation in NPR2. *Journal of Pediatric Endocrinology and Metabolism*, **2017**, 30, 111-116	1.6	9
87	Growth hormone deficiency with advanced bone age: phenotypic interaction between GHRH receptor and CYP21A2 mutations diagnosed by sanger and whole exome sequencing. *Archives of Endocrinology and Metabolism*, **2017**, 61, 633-636	2.2	3
86	Growth hormone insensitivity with immune dysfunction caused by a STAT5B mutation in the south of Brazil: evidence for a founder effect. *Genetics and Molecular Biology*, **2017**, 40, 436-441	2	2
85	Molecular analysis of brazilian patients with combined pituitary hormone deficiency and orthotopic posterior pituitary lobe reveals eight different PROP1 alterations with three novel mutations. *Clinical Endocrinology*, **2017**, 87, 725-732	3.4	9

84	Diagnosis and management of Silver-Russell syndrome: first international consensus statement. *Nature Reviews Endocrinology*, **2017**, 13, 105-124	15.2	224
83	Clinical Characterization of Patients With Autosomal Dominant Short Stature due to Aggrecan Mutations. *Journal of Clinical Endocrinology and Metabolism*, **2017**, 102, 460-469	5.6	68
82	Whole Exome Sequencing in the Investigation of Growth Disorders, Including Patients with Primary IGF-1 Deficiency. *Hormone Research in Paediatrics*, **2017**, 88, 421-422	3.3	1
81	Effects of Type 1 Insulin-Like Growth Factor Receptor Silencing in a Human Adrenocortical Cell Line. *Hormone and Metabolic Research*, **2016**, 48, 484-8	3.1	3
80	Partial correction of the dwarf phenotype by non-viral transfer of the growth hormone gene in mice: Treatment age is critical. *Growth Hormone and IGF Research*, **2016**, 26, 1-7	2	1
79	Sorafenib for the Treatment of Progressive Metastatic Medullary Thyroid Cancer: Efficacy and Safety Analysis. *Thyroid*, **2016**, 26, 414-9	6.2	30
78	A homozygous point mutation in the GH1 promoter (c.-223C>T) leads to reduced GH1 expression in siblings with isolated GH deficiency (IGHD). *European Journal of Endocrinology*, **2016**, 175, K7-K15	6.5	4
77	Two Patients with Severe Short Stature due to a FBN1 Mutation (p.Ala1728Val) with a Mild Form of Acromicric Dysplasia. *Hormone Research in Paediatrics*, **2016**, 86, 342-348	3.3	19
76	Frequent development of combined pituitary hormone deficiency in patients initially diagnosed as isolated growth hormone deficiency: a long term follow-up of patients from a single center. *Pituitary*, **2015**, 18, 561-7	4.3	23
75	STAT5B mutations in heterozygous state have negative impact on height: another clue in human stature heritability. *European Journal of Endocrinology*, **2015**, 173, 291-6	6.5	23
74	Influence of growth hormone receptor (GHR) exon 3 and -202A/C IGFBP-3 genetic polymorphisms on clinical and biochemical features and therapeutic outcome of patients with acromegaly. *Pituitary* , **2015**, 18, 666-73	4.3	10
73	Rare variants in SOS2 and LZTR1 are associated with Noonan syndrome. *Journal of Medical Genetics*, **2015**, 52, 413-21	5.8	144
72	Heterozygous NPR2 Mutations Cause Disproportionate Short Stature, Similar to Li-Weill Dyschondrosteosis. *Journal of Clinical Endocrinology and Metabolism*, **2015**, 100, E1133-42	5.6	47
71	Role of GLI2 in hypopituitarism phenotype. *Journal of Molecular Endocrinology*, **2015**, 54, R141-50	4.5	43
70	FGFR1 and PROKR2 rare variants found in patients with combined pituitary hormone deficiencies. *Endocrine Connections*, **2015**, 4, 100-7	3.5	28
69	Six additional cases of SEDC due to the same and recurrent R989C mutation in the COL2A1 gene--the clinical and radiological follow-up. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 894-901	2.5	7
68	Association Study of GWAS-Derived Loci with Height in Brazilian Children: Importance of MAP3K3, MMP24 and IGF1R Polymorphisms for Height Variation. *Hormone Research in Paediatrics*, **2015**, 84, 248-53	3.3	4
67	Further evidence of the importance of RIT1 in Noonan syndrome. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 2952-7	2.5	43

66	Autosomal recessive form of isolated growth hormone deficiency is more frequent than the autosomal dominant form in a Brazilian cohort. *Growth Hormone and IGF Research*, **2014**, 24, 180-6	2	5
65	Role of gonadotropin-releasing hormone receptor mutations in patients with a wide spectrum of pubertal delay. *Fertility and Sterility*, **2014**, 102, 838-846.e2	4.8	39
64	The growth hormone receptor exon 3 polymorphism is not associated with height or metabolic traits in healthy young adults. *Growth Hormone and IGF Research*, **2014**, 24, 123-9	2	9
63	Pathogenic mutations in GLI2 cause a specific phenotype that is distinct from holoprosencephaly. *Journal of Medical Genetics*, **2014**, 51, 413-8	5.8	45
62	A novel homologous model for gene therapy of dwarfism by non-viral transfer of the mouse growth hormone gene into immunocompetent dwarf mice. *Current Gene Therapy*, **2014**, 14, 44-51	4.3	6
61	Amplification of the insulin-like growth factor 1 receptor gene is a rare event in adrenocortical adenocarcinomas: searching for potential mechanisms of overexpression. *BioMed Research International*, **2014**, 2014, 936031	3	7
60	The E180splice mutation in the GHR gene causing Laron syndrome: witness of a Sephardic Jewish exodus from the Iberian Peninsula to the New World?. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1204-8	2.5	16
59	Role of the natriuretic peptide system in normal growth and growth disorders. *Hormone Research in Paediatrics*, **2014**, 82, 222-9	3.3	39
58	Genetic predictors of long-term response to growth hormone (GH) therapy in children with GH deficiency and Turner syndrome: the influence of a SOCS2 polymorphism. *Journal of Clinical Endocrinology and Metabolism*, **2014**, 99, E1808-13	5.6	14
57	Genome-wide screening of copy number variants in children born small for gestational age reveals several candidate genes involved in growth pathways. *European Journal of Endocrinology*, **2014**, 171, 253-62	6.5	26
56	Heterozygous mutations in natriuretic peptide receptor-B (NPR2) gene as a cause of short stature in patients initially classified as idiopathic short stature. *Journal of Clinical Endocrinology and Metabolism*, **2013**, 98, E1636-44	5.6	82
55	Differentiating the roles of STAT5B and STAT5A in human CD4+ T cells. *Clinical Immunology*, **2013**, 148, 227-36	9	35
54	Relatively high frequency of non-synonymous GLI2 variants in patients with congenital hypopituitarism without holoprosencephaly. *Clinical Endocrinology*, **2013**, 78, 551-7	3.4	28
53	Analysis of the insulin-like growth factor 1 receptor gene in children born small for gestational age: in vitro characterization of a novel mutation (p.Arg511Trp). *Clinical Endocrinology*, **2013**, 78, 558-63	3.4	12
52	The sitting height/height ratio for age in healthy and short individuals and its potential role in selecting short children for SHOX analysis. *Hormone Research in Paediatrics*, **2013**, 80, 449-56	3.3	41
51	The effect of fetal androgen metabolism-related gene variants on external genitalia virilization in congenital adrenal hyperplasia. *Clinical Genetics*, **2013**, 84, 482-8	4	13
50	Tegumentary manifestations of Noonan and Noonan-related syndromes. *Clinics*, **2013**, 68, 1079-83	2.3	8
49	Insulin-like growth factor 1 gene (CA)n repeats and a variable number of tandem repeats of the insulin gene in Brazilian children born small for gestational age. *Clinics*, **2013**, 68, 785-91	2.3	1

48	Growth standards of patients with Noonan and Noonan-like syndromes with mutations in the RAS/MAPK pathway. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2700-6	2.5	40
47	Incidental mild hyperglycemia in children: two MODY 2 families identified in Brazilian subjects. *Arquivos Brasileiros De Endocrinologia E Metabologia*, **2012**, 56, 519-24		7
46	Mutational analysis of TAC3 and TACR3 genes in patients with idiopathic central pubertal disorders. *Arquivos Brasileiros De Endocrinologia E Metabologia*, **2012**, 56, 646-52		36
45	Growth hormone pharmacogenetics: the interactive effect of a microsatellite in the IGF1 promoter region with the GHR-exon 3 and -202 A/C IGFBP3 variants on treatment outcomes of children with severe GH deficiency. *Pharmacogenomics Journal*, **2012**, 12, 439-45	3.5	18
44	KRAS gene mutations in Noonan syndrome familial cases cluster in the vicinity of the switch II region of the G-domain: report of another family with metopic craniosynostosis. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1178-84	2.5	13
43	Autoimmune disease and multiple autoantibodies in 42 patients with RASopathies. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1077-82	2.5	52
42	The role of fibroblast growth factor receptor 4 overexpression and gene amplification as prognostic markers in pediatric and adult adrenocortical tumors. *Endocrine-Related Cancer*, **2012**, 19, L11-3	5.7	16
41	Post-receptor IGF1 insensitivity restricted to the MAPK pathway in a Silver-Russell syndrome patient with hypomethylation at the imprinting control region on chromosome 11. *European Journal of Endocrinology*, **2012**, 166, 543-50	6.5	7
40	The interactive effect of GHR-exon 3 and -202 A/C IGFBP3 polymorphisms on rhGH responsiveness and treatment outcomes in patients with Turner syndrome. *Journal of Clinical Endocrinology and Metabolism*, **2012**, 97, E671-7	5.6	13
39	GH-releasing hormone receptor gene: a novel splice-disrupting mutation and study of founder effects. *Hormone Research in Paediatrics*, **2012**, 78, 165-72	3.3	16
38	Growth hormone receptor polymorphism and growth hormone therapy response in children: a Bayesian meta-analysis. *American Journal of Epidemiology*, **2012**, 175, 867-77	3.8	29
37	Growth responses following a single intra-muscular hGH plasmid administration compared to daily injections of hGH in dwarf mice. *Current Gene Therapy*, **2012**, 12, 437-43	4.3	3
36	Growth Defects in Noonan Syndrome **2012**, 2201-2215		
35	Pharmacogenetics of glucocorticoid replacement could optimize the treatment of congenital adrenal hyperplasia due to 21-hydroxylase deficiency. *Clinics*, **2011**, 66, 1361-6	2.3	7
34	Absence of GH-releasing hormone (GHRH) mutations in selected patients with isolated GH deficiency. *Journal of Clinical Endocrinology and Metabolism*, **2011**, 96, E1457-60	5.6	14
33	Novel inactivating mutations in the GH secretagogue receptor gene in patients with constitutional delay of growth and puberty. *European Journal of Endocrinology*, **2011**, 165, 233-41	6.5	42
32	Frequency of genetic polymorphisms of PXR gene in the Brazilian population. *Clinics*, **2011**, 66, 1041-4	2.3	7
31	A novel homozygous splice acceptor site mutation of KISS1R in two siblings with normosmic isolated hypogonadotropic hypogonadism. *European Journal of Endocrinology*, **2010**, 163, 29-34	6.5	47

30	A novel STAT5B mutation causing GH insensitivity syndrome associated with hyperprolactinemia and immune dysfunction in two male siblings. *European Journal of Endocrinology*, **2010**, 163, 349-55	6.5	42
29	Novel heterozygous nonsense GLI2 mutations in patients with hypopituitarism and ectopic posterior pituitary lobe without holoprosencephaly. *Journal of Clinical Endocrinology and Metabolism*, **2010**, 95, E384-91	5.6	83
28	PTPN11 and KRAS gene analysis in patients with Noonan and Noonan-like syndromes. *Genetic Testing and Molecular Biomarkers*, **2010**, 14, 425-32	1.6	23
27	Effectiveness of the combined recombinant human growth hormone and gonadotropin-releasing hormone analog therapy in pubertal patients with short stature due to SHOX deficiency. *Journal of Clinical Endocrinology and Metabolism*, **2010**, 95, 328-32	5.6	29
26	Steroidogenic factor 1 overexpression and gene amplification are more frequent in adrenocortical tumors from children than from adults. *Journal of Clinical Endocrinology and Metabolism*, **2010**, 95, 1458-62	5.6	50
25	Usefulness of MLPA in the detection of SHOX deletions. *European Journal of Medical Genetics*, **2010**, 53, 234-8	2.6	20
24	Co-occurring PTPN11 and SOS1 gene mutations in Noonan syndrome: does this predict a more severe phenotype?. *Arquivos Brasileiros De Endocrinologia E Metabologia*, **2010**, 54, 717-22		12
23	Short stature caused by isolated SHOX gene haploinsufficiency: update on the diagnosis and treatment. *Pediatric Endocrinology Reviews*, **2010**, 8, 79-85	1.1	7
22	Noonan syndrome and related disorders: a review of clinical features and mutations in genes of the RAS/MAPK pathway. *Hormone Research in Paediatrics*, **2009**, 71, 185-93	3.3	53
21	The -202 A allele of insulin-like growth factor binding protein-3 (IGFBP3) promoter polymorphism is associated with higher IGFBP-3 serum levels and better growth response to growth hormone treatment in patients with severe growth hormone deficiency. *Journal of Clinical Endocrinology and Metabolism*, **2009**, 94, 580-85	5.6	43
20	Comparison between weight-based and IGF-I-based growth hormone (GH) dosing in the treatment of children with GH deficiency and influence of exon 3 deleted GH receptor variant. *Growth Hormone and IGF Research*, **2009**, 19, 179-86	2	14
19	Growth hormone receptor exon 3 isoforms and their implication in growth disorders and treatment. *Hormone Research in Paediatrics*, **2009**, 71 Suppl 2, 55-63	3.3	10
18	Analysis of the PTPN11 gene in idiopathic short stature children and Noonan syndrome patients. *Clinical Endocrinology*, **2008**, 69, 426-31	3.4	7
17	High prevalence of pituitary magnetic resonance abnormalities and gene mutations in a cohort of Brazilian children with growth hormone deficiency and response to treatment. *Journal of Pediatric Endocrinology and Metabolism*, **2008**, 21, 673-80	1.6	8
16	Expression of insulin-like growth factor-II and its receptor in pediatric and adult adrenocortical tumors. *Journal of Clinical Endocrinology and Metabolism*, **2008**, 93, 3524-31	5.6	123
15	Cryptic intragenic deletion of the SHOX gene in a family with Leri-Weill dyschondrosteosis detected by Multiplex Ligation-Dependent Probe Amplification (MLPA). *Arquivos Brasileiros De Endocrinologia E Metabologia*, **2008**, 52, 1382-7		7
14	Exon 3-deleted genotype of growth hormone receptor (GHRd3) positively influences IGF-1 increase at generation test in children with idiopathic short stature. *Clinical Endocrinology*, **2007**, 67, 500-4	3.4	8
13	Polymorphisms identified in the upstream core polyadenylation signal of IGF1 gene exon 6 do not cause pre- and postnatal growth impairment. *Journal of Clinical Endocrinology and Metabolism*, **2007**, 92, 4889-92	5.6	20

12	Diagnosis and Long-Term Human Growth Hormone Treatment of a Boy with Noonan Syndrome. *Hormone Research in Paediatrics*, **2007**, 67, 98-101	3.3	
11	Muscular dystrophy-related quantitative and chemical changes in adenohypophysis GH-cells in golden retrievers. *Growth Hormone and IGF Research*, **2007**, 17, 480-91	2	18
10	SHOX mutations in idiopathic short stature and Leri-Weill dyschondrosteosis: frequency and phenotypic variability. *Clinical Endocrinology*, **2007**, 66, 130-5	3.4	47
9	Anthropometric evaluation of children with SHOX mutations can be used as indication for genetic studies in children of short stature. *Journal of Medical Genetics*, **2007**, 44, e90; author reply e91	5.8	
8	Growth hormone (GH) pharmacogenetics: influence of GH receptor exon 3 retention or deletion on first-year growth response and final height in patients with severe GH deficiency. *Journal of Clinical Endocrinology and Metabolism*, **2006**, 91, 1076-80	5.6	118
7	PTPN11 (protein tyrosine phosphatase, nonreceptor type 11) mutations and response to growth hormone therapy in children with Noonan syndrome. *Journal of Clinical Endocrinology and Metabolism*, **2005**, 90, 5156-60	5.6	66
6	Primary GH insensitivity (Laron syndrome) caused by a novel 4 kb deletion encompassing exon 5 of the GH receptor gene: effect of intermittent long-term treatment with recombinant human IGF-I. *European Journal of Endocrinology*, **2004**, 150, 635-42	6.5	15
5	The first homozygous mutation (S226I) in the highly-conserved WSXWS-like motif of the GH receptor causing Laron syndrome: supression of GH secretion by GnRH analogue therapy not restored by dihydrotestosterone administration. *Clinical Endocrinology*, **2004**, 60, 36-40	3.4	14
4	Acromegalic features in growth hormone (GH)-deficient patients after long-term GH therapy. *Clinical Endocrinology*, **2003**, 59, 788-92	3.4	12
3	Pituitary magnetic resonance imaging and function in patients with growth hormone deficiency with and without mutations in GHRH-R, GH-1, or PROP-1 genes. *Journal of Clinical Endocrinology and Metabolism*, **2002**, 87, 5076-84	5.6	104
2	An unusual phenotype of Frasier syndrome due to IVS9 +4C>T mutation in the WT1 gene: predominantly male ambiguous genitalia and absence of gonadal dysgenesis. *Journal of Clinical Endocrinology and Metabolism*, **2002**, 87, 2500-5	5.6	54
1	Developmental Syndromes of Ras/MAPK Pathway Dysregulation		2