James S Acierno

List of Publications by Year
in descending order

16 papers	1,381 citations	687363 13 h-index	996975 15 g-index
16 all docs	16 docs citations	16 times ranked	1377 citing authors

#	Article	IF	Citations
1	Digenic mutations account for variable phenotypes in idiopathic hypogonadotropic hypogonadism. Journal of Clinical Investigation, 2007, 117, 457-463.	8.2	338
2	Clinical Management of Congenital Hypogonadotropic Hypogonadism. Endocrine Reviews, 2019, 40, 669-710.	20.1	244
3	Mutations in fibroblast growth factor receptor 1 cause both Kallmann syndrome and normosmic idiopathic hypogonadotropic hypogonadism. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 6281-6286.	7.1	225
4	Mutations in fibroblast growth factor receptor 1 cause Kallmann syndrome with a wide spectrum of reproductive phenotypes. Molecular and Cellular Endocrinology, 2006, 254-255, 60-69.	3.2	176
5	Congenital hypogonadotropic hypogonadism and constitutional delay of growth and puberty have distinct genetic architectures. European Journal of Endocrinology, 2018, 178, 377-388.	3.7	95
6	<scp>KLB</scp> , encoding Î²â€Klotho, is mutated in patients with congenital hypogonadotropic hypogonadism. EMBO Molecular Medicine, 2017, 9, 1379-1397.	6.9	77
7	DCC/NTN1 complex mutations in patients with congenital hypogonadotropic hypogonadism impair GnRH neuron development. Human Molecular Genetics, 2018, 27, 359-372.	2.9	42
8	Î²-Klotho deficiency protects against obesity through a crosstalk between liver, microbiota, and brown adipose tissue. JCI Insight, 2017, 2, .	5.0	41
9	Neuron-Derived Neurotrophic Factor Is Mutated in Congenital Hypogonadotropic Hypogonadism. American Journal of Human Genetics, 2020, 106, 58-70.	6.2	39
10	Evaluating CHARGE syndrome in congenital hypogonadotropic hypogonadism patients harboring CHD7 variants. Genetics in Medicine, 2018, 20, 872-881.	2.4	38
11	Congenital Hypogonadotropic Hypogonadism with Anosmia and Gorlin Features Caused by a PTCH1 Mutation Reveals a New Candidate Gene for Kallmann Syndrome. Neuroendocrinology, 2021, 111, 99-114.	2.5	20
12	Integrating clinical and genetic approaches in the diagnosis of 46,XY disorders of sex development. Endocrine Connections, 2018, 7, 1480-1490.	1.9	18
13	The Reduction of Visceral Adipose Tissue after Roux-en-Y Gastric Bypass Is more Pronounced in Patients with Impaired Glucose Metabolism. Obesity Surgery, 2018, 28, 4006-4013.	2.1	16
14	Pathogenic mosaic variants in congenital hypogonadotropic hypogonadism. Genetics in Medicine, 2020, 22, 1759-1767.	2.4	7
15	A novel CHD7 mutation in an adolescent presenting with growth and pubertal delay. Annals of Pediatric Endocrinology and Metabolism, 2019, 24, 49-54.	2.3	5
16	Testosterone-induced increase in libido in a patient with a loss-of-function mutation in the AR gene. Endocrinology, Diabetes and Metabolism Case Reports, 2021, 2021, .	0.5	0