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List of Publications by Year in descending order

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12
papers

940
citations

680236

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971612

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14
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1715
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations and variants of ONECUT1 in diabetes. <i>Nature Medicine</i> , 2021, 27, 1928-1940.	29.9	27
2	YIPF5 mutations cause neonatal diabetes and microcephaly through endoplasmic reticulum stress. <i>Journal of Clinical Investigation</i> , 2020, 130, 6338-6353.	6.6	63
3	dUTPase (<i>DUT</i>) Is Mutated in a Novel Monogenic Syndrome With Diabetes and Bone Marrow Failure. <i>Diabetes</i> , 2017, 66, 1086-1096.	0.9	23
4	Juvenile-Onset Diabetes and Congenital Cataract: "Double-Gene" Mutations Mimicking a Syndromic Diabetes Presentation. <i>Genes</i> , 2017, 8, 309.	2.4	8
5	Microscopic and ultrastructural features in Wolcott-Rallison syndrome, a permanent neonatal diabetes mellitus: about two autopsy cases. <i>Pediatric Diabetes</i> , 2015, 16, 510-520.	3.0	13
6	A Missense Mutation in <i>PPP1R15B</i> Causes a Syndrome Including Diabetes, Short Stature, and Microcephaly. <i>Diabetes</i> , 2015, 64, 3951-3962.	0.9	80
7	A novel <i>ALMS1</i> splice mutation in a non-obese juvenile-onset insulin-dependent syndromic diabetic patient. <i>European Journal of Human Genetics</i> , 2014, 22, 140-143.	2.9	20
8	Wolcott-Rallison syndrome due to the same mutation (W522X) in <i>EIF2AK3</i> in two unrelated families and review of the literature*. <i>Pediatric Diabetes</i> , 2010, 11, 279-285.	3.0	44
9	A Novel Hypomorphic <i>PDX1</i> Mutation Responsible for Permanent Neonatal Diabetes With Subclinical Exocrine Deficiency. <i>Diabetes</i> , 2010, 59, 733-740.	0.9	86
10	Severe FOXP3+ and Naïve T Lymphopenia in a Non-IPEX Form of Autoimmune Enteropathy Combined With an Immunodeficiency. <i>Gastroenterology</i> , 2007, 132, 1694-1704.	1.4	28
11	Mutations in <i>GLIS3</i> are responsible for a rare syndrome with neonatal diabetes mellitus and congenital hypothyroidism. <i>Nature Genetics</i> , 2006, 38, 682-687.	20.2	335
12	Crohn's disease associated <i>CARD15</i> (<i>NOD2</i>) variants are not involved in the susceptibility to type 1 diabetes. <i>Molecular Genetics and Metabolism</i> , 2005, 86, 379-383.	2.2	12