

List of Publications by Year in descending order

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1696
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#	ARTICLE	IF	CITATIONS
1	Disruption of ALX1 Causes Extreme Microphthalmia and Severe Facial Clefing: Expanding the Spectrum of Autosomal-Recessive ALX-Related Frontonasal Dysplasia. American Journal of Human Genetics, 2010, 86, 789-796.	6.2	128
2	ALX4 dysfunction disrupts craniofacial and epidermal development. Human Molecular Genetics, 2009, 18, 4357-4366.	2.9	103
3	Evidence from autoimmune thyroiditis of skewed X-chromosome inactivation in female predisposition to autoimmunity. European Journal of Human Genetics, 2006, 14, 791-797.	2.8	99
4	Analysis of skewed X-chromosome inactivation in females with rheumatoid arthritis and autoimmune thyroid diseases. Arthritis Research and Therapy, 2009, 11, R106.	3.5	90
5	Mutations in the very low-density lipoprotein receptor <i>VLDLR</i> cause cerebellar hypoplasia and quadrupedal locomotion in humans. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 4232-4236.	7.1	88
6	STK4 (MST1) deficiency in two siblings with autoimmune cytopenias: A novel mutation. Clinical Immunology, 2015, 161, 316-323.	3.2	73
7	Mutations in the interleukin receptor <i>IL</i>1<i>RA</i> cause autosomal recessive Crouzon-like craniosynostosis. Molecular Genetics & Genomic Medicine, 2013, 1, 223-237.	1.2	70
8	Mutations in IRX5 impair craniofacial development and germ cell migration via SDF1. Nature Genetics, 2012, 44, 709-713.	21.4	68
9	Skewed X-Chromosome Inactivation in Scleroderma. Clinical Reviews in Allergy and Immunology, 2008, 34, 352-355.	6.5	46
10	Increased frequency of extremely skewed X chromosome inactivation in juvenile idiopathic arthritis. Arthritis and Rheumatism, 2009, 60, 3410-3412.	6.7	23
11	Loss-of-Function Mutations in ELMO2 Cause Intraosseous Vascular Malformation by Impeding RAC1 Signaling. American Journal of Human Genetics, 2016, 99, 299-317.	6.2	23
12	Reply to Herz <i>et al.</i> and Humphrey <i>et al.</i> : Genetic heterogeneity of cerebellar hypoplasia with quadrupedal locomotion. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, E32-3.	7.1	9
13	Intracerebroventricularly injected nesfatin-1 activates central cyclooxygenase and lipoxigenase pathways. Autonomic Neuroscience: Basic and Clinical, 2020, 226, 102670.	2.8	4
14	Extremely skewed X-chromosome inactivation is increased in pre-eclampsia. Human Genetics, 2007, 121, 101-105.	3.8	3