Boris Utsch

List of Publications by Year
in descending order

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27 papers	2,002 citations	361413 20 h-index	526287 27 g-index
28 all docs	28 docs citations	28 times ranked	2087 citing authors

#	Article	IF	Citations
1	The centrosomal protein nephrocystin-6 is mutated in Joubert syndrome and activates transcription factor ATF4. Nature Genetics, 2006, 38, 674-681.	21.4	535
2	Nephrocystin-5, a ciliary IQ domain protein, is mutated in Senior-Loken syndrome and interacts with RPGR and calmodulin. Nature Genetics, 2005, 37, 282-288.	21.4	367
3	Evidence of Oligogenic Inheritance in Nephronophthisis. Journal of the American Society of Nephrology: JASN, 2007, 18, 2789-2795.	6.1	141
4	Mutation analysis of NPHP6/CEP290 in patients with Joubert syndrome and Senior Loken syndrome. Journal of Medical Genetics, 2007, 44, 657-663.	3.2	93
5	Identification of the first AHI1 gene mutations in nephronophthisis-associated Joubert syndrome. Pediatric Nephrology, 2006, 21, 32-35.	1.7	87
6	A novel stable polyalanine [poly(A)] expansion in the HOXA13 gene associated with hand-foot-genital syndrome: proper function of poly(A)-harbouring transcription factors depends on a critical repeat length?. Human Genetics, 2002, 110, 488-494.	3.8	75
7	Novel OCRL1 Mutations in Patients With the Phenotype of Dent Disease. American Journal of Kidney Diseases, 2006, 48, 942.e1-942.e14.	1.9	68
8	Mutational analysis of theNPHP4 gene in 250 patients with nephronophthisis. Human Mutation, 2005, 25, 411-411.	2.5	60
9	Functional evaluation of Dentâ€™s disease-causing mutations: implications for ClC-5 channel trafficking and internalization. Human Genetics, 2005, 117, 228-237.	3.8	52
10	Recent advances in understanding the clinical and genetic heterogeneity of Dent's disease. Nephrology Dialysis Transplantation, 2006, 21, 2708-2717.	0.7	51
11	Novel NCCT Gene Mutations as a Cause of Gitelmanâ€™s Syndrome and a Systematic Review of Mutant and Polymorphic NCCT Alleles. Kidney and Blood Pressure Research, 2002, 25, 354-362.	2.0	47
12	Polyalanine expansion in HOXA13: three new affected families and the molecular consequences in a mouse model. Human Molecular Genetics, 2004, 13, 2841-2851.	2.9	47
13	Hypercalciuria in patients with CLCN5 mutations. Pediatric Nephrology, 2006, 21, 1241-1250.	1.7	45
14	A Hospital-Based Intermittent Nocturnal Hemodialysis Program for Children and Adolescents. Journal of Pediatrics, 2011, 158, 95-99.e1.	1.8	43
15	Haplotype analysis improves molecular diagnostics of autosomal recessive polycystic kidney disease. American Journal of Kidney Diseases, 2005, 45, 77-87.	1.9	41
16	Genome-wide Association Study and Meta-Analysis Identify ISL1 as Genome-wide Significant Susceptibility Gene for Bladder Exstrophy. PLoS Genetics, 2015, 11, e1005024.	3.5	41
17	Delayed transport of tissue-nonspecific alkaline phosphatase with missense mutations causing hypophosphatasia. European Journal of Medical Genetics, 2007, 50, 367-378.	1.3	34
18	Phenotype Severity in the Bladder Exstrophy-Epispadias Complex: Analysis of Genetic and Nongenetic Contributing Factors in 441 Families from North America and Europe. Journal of Pediatrics, 2011, 159, 825-831.e1.	1.8	33

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19	Mapping a new suggestive gene locus for autosomal dominant nephrolithiasis to chromosome 9q33.2â€“q34.2 by total genome search for linkage. Nephrology Dialysis Transplantation, 2005, 20, 909-914.	0.7	26
20	Molecular characterization of HOXA13 polyalanine expansion proteins in handâ€“footâ€“genital syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 3161-3168.	1.2	25
21	Genome-wide association study and mouse expression data identify a highly conserved 32 kb intergenic region between WNT3 and WNT9b as possible susceptibility locus for isolated classic exstrophy of the bladder. Human Molecular Genetics, 2014, 23, 5536-5544.	2.9	19
22	Immature Gastric Teratoma of the Lesser Curvature in a Male Infant. Journal of Pediatric Gastroenterology and Nutrition, 2001, 32, 204-206.	1.8	15
23	Four Additional CLCN5 Exons Encode a Widely Expressed Novel Long CLC-5 Isoform but Fail to Explain Dentâ€™s Phenotype in Patients without Mutations in the Short Variant. Kidney and Blood Pressure Research, 2003, 26, 176-184.	2.0	15
24	Exclusion of WTAP and HOXA13 as candidate genes for isolated hypospadias. Scandinavian Journal of Urology and Nephrology, 2003, 37, 498-501.	1.4	13
25	Dent diseaseâ€like phenotype and the chloride channel ClCâ€4 (CLCN4) gene. American Journal of Medical Genetics Part A, 2004, 128A, 434-435.	2.4	13
26	Bladder exstrophy and Epstein type congenital macrothrombocytopenia: Evidence for a common cause?. American Journal of Medical Genetics, Part A, 2006, 140A, 2251-2253.	1.2	11
27	Homozygous ?-thalassemia associated with hypospadias: SEA-type deletion does not affect expression of the -14 gene and loss of the ?1-globin gene on 16p13.3 is compensated by its duplicate ?2 on chromosome 10. American Journal of Medical Genetics Part A, 2001, 101, 286-287.	2.4	5