Heiko Martin Reutter

List of Publications by Year
in descending order

157	4,832	126907	118850
papers	citations	33 h-index	62 g-index

157	157	157	5105
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Isolated cytokineâ€enriched pericardial effusion: A likely key feature for <scp>AymÃ©â€Gripp</scp> syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 624-627.	1.2	2
2	First genome-wide association study of esophageal atresia identifies three genetic risk loci at CTNNA3, FOXF1/FOXC2/FOXL1, and HNF1B. Human Genetics and Genomics Advances, 2022, 3, 100093.	1.7	4
3	Resequencing of <scp>VEGFR3</scp> pathway genes implicate <scp>GJC2</scp> and <scp>FLT4</scp> in the formation of primary congenital chylothorax. American Journal of Medical Genetics, Part A, 2022, 188, 1607-1611.	1.2	3
4	Nonurgent Visits to the Pediatric Emergency Department before and during the First Peak of the COVID-19 Pandemic. International Journal of Pediatrics (United Kingdom), 2022, 2022, 1-7.	0.8	1
5	Reâ€sequencing of candidate genes <scp>FOXF1</scp>, <scp>HSPA6</scp>, <scp>HAAO</scp>, and <scp>KYNU</scp> in 522 individuals with <scp>VATER</scp>/<scp>VACTERL</scp>, <scp>VACTER</scp>/<scp>VACTERL</scp>â€like association, and isolated anorectal malformation. Birth Defects Research, 2022, 114, 478-486.	1.5	6
6	Exome sequencing in individuals with cardiovascular laterality defects identifies potential candidate genes. European Journal of Human Genetics, 2022, , .	2.8	1
7	Definition, diagnosis and clinical management of non-obstructive kidney dysplasia: a consensus statement by the ERKNet Working Group on Kidney Malformations. Nephrology Dialysis Transplantation, 2022, 37, 2351-2362.	0.7	6
8	Currarino syndrome: a comprehensive genetic review of a rare congenital disorder. Orphanet Journal of Rare Diseases, 2021, 16, 167.	2.7	10
9	ANKRD11 variants: <scp>KBG</scp> syndrome and beyond. Clinical Genetics, 2021, 100, 187-200.	2.0	21
10	Biallelic and monoallelic variants in PLXNA1 are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. Genetics in Medicine, 2021, 23, 1715-1725.	2.4	22
11	Successful ECMO therapy in a child with COVIDâ€19â€associated ARDS and acute lymphoblastic leukemia. Pediatric Blood and Cancer, 2021, 68, e29100.	1.5	4
12	Parental risk factors for congenital diaphragmatic hernia â€“ a large German case-control study. BMC Pediatrics, 2021, 21, 278.	1.7	13
13	The Genomic Architecture of Bladder Exstrophy Epispadias Complex. Genes, 2021, 12, 1149.	2.4	8
14	The Role of De Novo Variants in Formation of Human Anorectal Malformations. Genes, 2021, 12, 1298.	2.4	1
15	Exome survey of individuals affected by VATER / VACTERL with renal phenotypes identifies phenocopies and novel candidate genes. American Journal of Medical Genetics, Part A, 2021, 185, 3784-3792.	1.2	6
16	Genome-Wide Survey for Microdeletions or -Duplications in 155 Patients with Lower Urinary Tract Obstructions (LUTO). Genes, 2021, 12, 1449.	2.4	4
17	The Role of De Novo Variants in Patients with Congenital Diaphragmatic Hernia. Genes, 2021, 12, 1405.	2.4	5
18	A Prevalence Estimation of Exstrophy and Epispadias in Germany From Public Health Insurance Data. Frontiers in Pediatrics, 2021, 9, 648414.	1.9	4

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19	Unraveling the Genetics of Congenital Diaphragmatic Hernia: An Ongoing Challenge. Frontiers in Pediatrics, 2021, 9, 800915.	1.9	12
20	Genetic Counseling for Birth Defects. European Journal of Pediatric Surgery, 2021, 31, 467-467.	1.3	0
21	Genetic Counseling for Bladder Exstrophy-Epispadias Complex. European Journal of Pediatric Surgery, 2021, 31, 468-471.	1.3	1
22	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	10.3	43
23	SLC20A1 Is Involved in Urinary Tract and Urorectal Development. Frontiers in Cell and Developmental Biology, 2020, 8, 567.	3.7	22
24	Lessons Learned from CNV Analysis of Major Birth Defects. International Journal of Molecular Sciences, 2020, 21, 8247.	4.1	8
25	Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. American Journal of Human Genetics, 2020, 107, 727-742.	6.2	25
26	Human exome and mouse embryonic expression data implicate ZFHX3, TRPS1, and CHD7 in human esophageal atresia. PLoS ONE, 2020, 15, e0234246.	2.5	9
27	Treatment Strategies and Outcome of the Exstrophyâ€“Epispadias Complex in Germany: Data From the German CURE-Net. Frontiers in Pediatrics, 2020, 8, 174.	1.9	6
28	A Genetics-First Approach Revealed Monogenic Disorders in Patients With ARM and VACTERL Anomalies. Frontiers in Pediatrics, 2020, 8, 310.	1.9	17
29	DNA Methylation and Bladder Cancer: Where Genotype does not Predict Phenotype. Current Genomics, 2020, 21, 34-36.	1.6	17
30	Title is missing!. , 2020, 15, e0234246.		0
31	Title is missing!. , 2020, 15, e0234246.		0
32	Title is missing!. , 2020, 15, e0234246.		0
33	Title is missing!. , 2020, 15, e0234246.		0
34	Title is missing!. , 2020, 15, e0234246.		0
35	Title is missing!. , 2020, 15, e0234246.		0
36	Association Between Exstrophy-epispadias Complex And Congenital Anomalies: A German Multicenter Study. Urology, 2019, 123, 210-220.	1.0	9

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37	Male infant with paternal uniparental diploidy mosaicism and a 46,XX/46,XY karyotype. American Journal of Medical Genetics, Part A, 2019, 179, 2252-2256.	1.2	7
38	Classic bladder exstrophy and adenocarcinoma of the bladder: Methylome analysis provide no evidence for underlying disease-mechanisms of this association. Cancer Genetics, 2019, 235-236, 18-20.	0.4	10
39	Exome chip association study excluded the involvement of rare coding variants with large effect sizes in the etiology of anorectal malformations. PLoS ONE, 2019, 14, e0217477.	2.5	3
40	Rare Variants in BNC2 Are Implicated in Autosomal-Dominant Congenital Lower Urinary-Tract Obstruction. American Journal of Human Genetics, 2019, 104, 994-1006.	6.2	47
41	HSPA6: A new autosomal recessive candidate gene for the VATER/VACTERL malformation spectrum. Birth Defects Research, 2019, 111, 591-597.	1.5	15
42	AUTHOR REPLY. Urology, 2019, 123, 219-220.	1.0	0
43	Gastrointestinal diseases among relatives of patients with esophageal atresia with or without tracheoesophageal fistula. Translational Pediatrics, 2019, 8, 378-382.	1.2	0
44	CAKUT and Autonomic Dysfunction Caused by Acetylcholine Receptor Mutations. American Journal of Human Genetics, 2019, 105, 1286-1293.	6.2	18
45	A classic twin study of lower urinary tract obstruction: Report of 3 cases and literature review. LUTS: Lower Urinary Tract Symptoms, 2019, 11, O85-O88.	1.3	6
46	Expanding the knowledge on development of CAKUT: molecular genetics and beyond. Annals of Translational Medicine, 2019, 7, 596-596.	1.7	0
47	Nonsyndromic cleft palate: An association study at GWAS candidate loci in a multiethnic sample. Birth Defects Research, 2018, 110, 871-882.	1.5	11
48	Exome sequencing in syndromic brain malformations identifies novel mutations in ACTB, and SLC9A6, and suggests BAZ1A as a new candidate gene. Birth Defects Research, 2018, 110, 587-597.	1.5	21
49	Esophageal Atresia with or without Tracheoesophageal Fistula (EA/TEF): Association of Different EA/TEF Subtypes with Specific Co-occurring Congenital Anomalies and Implications for Diagnostic Workup. European Journal of Pediatric Surgery, 2018, 28, 176-182.	1.3	14
50	An examination of the factors affecting intestinal wall integrity in newborns at birth. Journal of Maternal-Fetal and Neonatal Medicine, 2018, 31, 294-299.	1.5	0
51	Early postnatal echocardiographic assessment of pulmonary blood flow in newborns with congenital diaphragmatic hernia. Journal of Perinatal Medicine, 2018, 46, 735-743.	1.4	16
52	Sexual Function and Quality of Life in Adult Male Individuals with Exstrophy-Epispadias Complexâ€"a Survey of the German CURE-Network. Urology, 2018, 112, 215-221.	1.0	9
53	Towards a Central Role of ISL1 in the Bladder Exstrophyâ€"Epispadias Complex (BEEC): Computational Characterization of Genetic Variants and Structural Modelling. Genes, 2018, 9, 609.	2.4	6
54	Congenital intrahepatic portocaval shunts and hypoglycemia due to secondary hyperinsulinism: a case report and review of the literature. Journal of Medical Case Reports, 2018, 12, 336.	0.8	7

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55	Needs Assessment in Care of Adults With Anorectal Malformations and Exstrophy-Epispadias Complex in Germany. Frontiers in Pediatrics, 2018, 6, 392.	1.9	2
56	De Novo Duplication of 11p15 Associated With Congenital Diaphragmatic Hernia. Frontiers in Pediatrics, 2018, 6, 116.	1.9	2
57	Circulating microRNAs are associated with Pulmonary Hypertension and Development of Chronic Lung Disease in Congenital Diaphragmatic Hernia. Scientific Reports, 2018, 8, 10735.	3.3	34
58	Clinical and echocardiographic risk factors for extubation failure in infants with congenital diaphragmatic hernia. Paediatric Anaesthesia, 2018, 28, 864-872.	1.1	5
59	Extracorporeal membrane oxygenation support in a newborn with lower urinary tract obstruction and pulmonary hypoplasia: a case report. Journal of Medical Case Reports, 2018, 12, 210.	0.8	2
60	Whole-Exome Sequencing Identifies Causative Mutations in Families with Congenital Anomalies of the Kidney and Urinary Tract. Journal of the American Society of Nephrology: JASN, 2018, 29, 2348-2361.	6.1	147
61	Low maternal folate concentrations and maternal MTHFR C677T polymorphism are associated with an increased risk for neural tube defects in offspring: a case-control study among Pakistani case and control mothers. Asia Pacific Journal of Clinical Nutrition, 2018, 27, 253-260.	0.4	8
62	Evaluation of two commercially available ELISA kits for the determination of melatonin concentrations in amniotic fluid throughout pregnancy. Annals of Clinical Biochemistry, 2017, 54, 107-112.	1.6	11
63	Quality of Life after Surgical Treatment for Esophageal Atresia: Long-Term Outcome of 154 Patients. European Journal of Pediatric Surgery, 2017, 27, 443-448.	1.3	19
64	ISL1 is a major susceptibility gene for classic bladder exstrophy and a regulator of urinary tract development. Scientific Reports, 2017, 7, 42170.	3.3	41
65	Role of the LF-SINEâ€"Derived Distal ISL1 Enhancer in Patients with Classic Bladder Exstrophy. Journal of Pediatric Genetics, 2017, 06, 169-173.	0.7	3
66	Arrayâ€based molecular karyotyping in 115 VATER/VACTERL and VATER/VACTERLâ€like patients identifies diseaseâ€causing copy number variations. Birth Defects Research, 2017, 109, 1063-1069.	1.5	26
67	Targeted Resequencing of Putative Growth-Related Genes Using Whole Exome Sequencing in Patients with Severe Primary IGF-I Deficiency. Hormone Research in Paediatrics, 2017, 88, 408-417.	1.8	9
68	Overlapping SETBP1 gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies. PLoS Genetics, 2017, 13, e1006683.	3.5	35
69	Epidemiologic analysis of families with isolated anorectal malformations suggests high prevalence of autosomal dominant inheritance. Orphanet Journal of Rare Diseases, 2017, 12, 180.	2.7	10
70	Parental risk factors of anorectal malformations: Analysis with a regional populationâ€based control group. Birth Defects Research Part A: Clinical and Molecular Teratology, 2016, 106, 133-141.	1.6	13
71	PLAGL1 epimutation and bladder exstrophy: Coincidence or concurrent etiology?. Birth Defects Research Part A: Clinical and Molecular Teratology, 2016, 106, 724-728.	1.6	2
72	Targeted sequencing of 96 renal developmental microRNAs in 1213 individuals from 980 families with congenital anomalies of the kidney and urinary tract. Nephrology Dialysis Transplantation, 2016, 31, 1280-1283.	0.7	15

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73	Whole-Exome Sequencing in Nine Monozygotic Discordant Twins. Twin Research and Human Genetics, 2016, 19, 60-65.	0.6	24
74	Arrayâ€based molecular karyotyping in fetal brain malformations: Identification of novel candidate genes and chromosomal regions. Birth Defects Research Part A: Clinical and Molecular Teratology, 2016, 106, 16-26.	1.6	13
75	More than fetal urine: enteral uptake of amniotic fluid as a major predictor for fetal growth during late gestation. European Journal of Pediatrics, 2016, 175, 825-831.	2.7	14
76	Fulminant Pneumococcal Meningoencephalitis and Widespread Cerebritis with Multiple Infarctions Caused by Nonâ€"PCV13-Serotype 23A in a 12-Month-Old Girl with Down Syndrome. Journal of Pediatric Infectious Diseases, 2016, 10, 085-088.	0.2	0
77	Melatonin Concentrations and Antioxidative Capacity of Human Breast Milk According to Gestational Age and the Time of Day. Journal of Human Lactation, 2016, 32, NP105-NP110.	1.6	45
78	Evaluation of sexual function in females with exstrophy-epispadias-complex: a survey of the multicenter German CURE-Net. Journal of Pediatric Urology, 2016, 13, 183.e1-183.e6.	1.1	7
79	Copy number variations in 375 patients with oesophageal atresia and/or tracheoesophageal fistula. European Journal of Human Genetics, 2016, 24, 1715-1723.	2.8	27
80	Loss of Function of GALNT2 Lowers High-Density Lipoproteins in Humans, Nonhuman Primates, and Rodents. Cell Metabolism, 2016, 24, 234-245.	16.2	103
81	Array-based molecular karyotyping in fetuses with isolated brain malformations identifies disease-causing CNVs. Journal of Neurodevelopmental Disorders, 2016, 8, 11.	3.1	24
82	CNV analysis in 169 patients with bladder exstrophy-epispadias complex. BMC Medical Genetics, 2016, 17, 35.	2.1	15
83	Familial tetrasomy 4q35.2 associated with congenital diaphragmatic hernia and unilateral renal agenesis: a case report. Journal of Medical Case Reports, 2016, 10, 76.	0.8	1
84	Sequencing the GRHL3 Coding Region Reveals Rare Truncating Mutations and a Common Susceptibility Variant for Nonsyndromic Cleft Palate. American Journal of Human Genetics, 2016, 98, 755-762.	6.2	92
85	Underlying genetic factors of the VATER/VACTERL association with special emphasis on the â€œRenalâ€• phenotype. Pediatric Nephrology, 2016, 31, 2025-2033.	1.7	34
86	Infancy-Onset T1DM, Short Stature, and Severe Immunodysregulation in Two Siblings With a Homozygous LRBA Mutation. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 898-904.	3.6	43
87	The Challenges of the European Anorectal Malformations-Net Registry. European Journal of Pediatric Surgery, 2015, 25, 481-487.	1.3	22
88	Lowe syndrome/Dent-2 disease: A comprehensive review of known and novel aspects. Journal of Pediatric Genetics, 2015, 02, 053-068.	0.7	22
89	Targeted Resequencing of 29 Candidate Genes and Mouse Expression Studies ImplicateZIC3andFOXF1in Human VATER/VACTERL Association. Human Mutation, 2015, 36, 1150-1154.	2.5	46
90	Mutations in PTF1A are not a common cause for human VATER/VACTERL association or neural tube defects mirroring Danforthâ€™s short tail mouse. Molecular Medicine Reports, 2015, 12, 1579-1583.	2.4	3

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91	Genetics of Bladder-Exstrophy-Epispadias Complex (BEEC): Systematic Elucidation of Mendelian and Multifactorial Phenotypes. Current Genomics, 2015, 17, 4-13.	1.6	36
92	Scimitar syndrome in a case with VACTERL association. Cardiology in the Young, 2015, 25, 606-609.	0.8	6
93	WNT3 involvement in human bladder exstrophy and cloaca development in zebrafish. Human Molecular Genetics, 2015, 24, 5069-5078.	2.9	23
94	Antioxidative status and oxidative stress in the fetal circulation at birth: the effects of time of delivery and presence of labor. Early Human Development, 2015, 91, 119-124.	1.8	12
95	Mutations in TBX18 Cause Dominant Urinary Tract Malformations via Transcriptional Dysregulation of Ureter Development. American Journal of Human Genetics, 2015, 97, 291-301.	6.2	72
96	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
97	Genome-wide array data and next generation sequencing unravel the etiology of urogenital malformations. Journal of Pediatric Genetics, 2015, 01, 209-216.	0.7	1
98	Mutations of the SLIT2â€“ROBO2 pathway genes SLIT2 and SRGAP1 confer risk for congenital anomalies of the kidney and urinary tract. Human Genetics, 2015, 134, 905-916.	3.8	62
99	Sexual function in adult patients with classic bladder exstrophy: A multicenter study. Journal of Pediatric Urology, 2015, 11, 125.e1-125.e6.	1.1	30
100	Genomeâ€wide mapping of copy number variations in patients with both anorectal malformations and central nervous system abnormalities. Birth Defects Research Part A: Clinical and Molecular Teratology, 2015, 103, 235-242.	1.6	18
101	Mild Recessive Mutations in Six Fraser Syndromeâ€“Related Genes Cause Isolated Congenital Anomalies of the Kidney and Urinary Tract. Journal of the American Society of Nephrology: JASN, 2014, 25, 1917-1922.	6.1	97
102	Evidence for annular pancreas as an associated anomaly in the VATER/VACTERL association and investigation of the gene encoding pancreas specific transcription factor 1A as a candidate gene. American Journal of Medical Genetics, Part A, 2014, 164, 1611-1613.	1.2	10
103	Mutations in 12 known dominant disease-causing genes clarify many congenital anomalies of the kidney and urinary tract. Kidney International, 2014, 85, 1429-1433.	5.2	203
104	Classic bladder exstrophy: Frequent 22q11.21 duplications and definition of a 414 kb phenocritical region. Birth Defects Research Part A: Clinical and Molecular Teratology, 2014, 100, 512-517.	1.6	21
105	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
106	Whole-exome resequencing reveals recessive mutations in TRAP1 in individuals with CAKUT and VACTERL association. Kidney International, 2014, 85, 1310-1317.	5.2	106
107	An Approach to the Identification of Anomalies and Etiologies in Neonates with Identified or Suspected VACTERL (Vertebral Defects, Anal Atresia, Tracheo-Esophageal Fistula with Esophageal) Tj ETQq1 1 0.784314 rgBT /Overlock 2014, 164, 451-457.e1.	1.8	83
108	Heterozygous FGF8 mutations in patients presenting cryptorchidism and multiple VATER/VACTERL features without limb anomalies. Birth Defects Research Part A: Clinical and Molecular Teratology, 2014, 100, 750-759.	1.6	14

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109	De novo microduplications at 1q41, 2q37.3, and 8q24.3 in patients with VATER/VACTERL association. European Journal of Human Genetics, 2013, 21, 1377-1382.	2.8	38
110	Candidate gene association study implicates p63 in the etiology of nonsyndromic bladderâ€exstrophyâ€epispadias complex. Birth Defects Research Part A: Clinical and Molecular Teratology, 2013, 97, 759-763.	1.6	8
111	De novo 13q deletions in two patients with mild anorectal malformations as part of VATER/VACTERL and VATER/VACTERLâ€like association and analysis of EFNB2 in patients with anorectal malformations. American Journal of Medical Genetics, Part A, 2013, 161, 3035-3041.	1.2	32
112	Isolated bladder exstrophy associated with a de novo 0.9 Mb microduplication on chromosome 19p13.12. Birth Defects Research Part A: Clinical and Molecular Teratology, 2013, 97, 133-139.	1.6	18
113	Yunis-VarÃ³n Syndrome Is Caused by Mutations in FIG4, Encoding a Phosphoinositide Phosphatase. American Journal of Human Genetics, 2013, 92, 781-791.	6.2	124
114	Assisted Reproductive Techniques and Risk of Exstrophy-Epispadias Complex: A German Case-Control Study. Journal of Urology, 2013, 189, 1524-1529.	0.4	22
115	CNV Analysis in Monozygotic Twin Pairs Discordant for Urorectal Malformations. Twin Research and Human Genetics, 2013, 16, 802-807.	0.6	9
116	Second study on the recurrence risk of isolated esophageal atresia with or without tracheaâ€esophageal fistula among firstâ€degree relatives: No evidence for increased risk of recurrence of EA/TEF or for malformations of the VATER/VACTERL association spectrum. Birth Defects Research Part A: Clinical and Molecular Teratology, 2013, 97, 786-791.	1.6	12
117	VATER/VACTERL association. Clinical Dysmorphology, 2012, 21, 191-195.	0.3	26
118	A Classic Twin Study of Isolated Gastroschisis. Fetal and Pediatric Pathology, 2012, 31, 324-330.	0.7	10
119	Involvement of the WNT and FGF signaling pathways in non-isolated anorectal malformations: Sequencing analysis of WNT3A, WNT5A, WNT11, DACT1, FGF10, FGFR2 and the T gene. International Journal of Molecular Medicine, 2012, 30, 1459-1464.	4.0	24
120	Clinical geneticists' views of VACTERL/VATER association. American Journal of Medical Genetics, Part A, 2012, 158A, 3087-3100.	1.2	78
121	Primary pulmonary hypertension, congenital heart defect, central nervous system malformations, hypo- and aplastic toes: Another case of Yunis-VarÃ³n syndrome or report of a new entity. European Journal of Medical Genetics, 2012, 55, 27-31.	1.3	5
122	Genome-wide meta-analyses of nonsyndromic cleft lip with or without cleft palate identify six new risk loci. Nature Genetics, 2012, 44, 968-971.	21.4	311
123	Nine new twin pairs with esophageal atresia: A review of the literature and performance of a twin study of the disorder. Birth Defects Research Part A: Clinical and Molecular Teratology, 2012, 94, 182-186.	1.6	17
124	Familial occurrence of the VATER/VACTERL association. Pediatric Surgery International, 2012, 28, 725-729.	1.4	36
125	Inheritance of the VATER/VACTERL association. Pediatric Surgery International, 2012, 28, 681-685.	1.4	29
126	De novo microduplication at 22q11.21 in a patient with VACTERL association. European Journal of Medical Genetics, 2011, 54, 9-13.	1.3	56

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127	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
128	Muscarinic Acetylcholine Receptor M3 Mutation Causes Urinary Bladder Disease and a Prune-Belly-like Syndrome. American Journal of Human Genetics, 2011, 89, 668-674.	6.2	89
129	Autosomal-dominant non-syndromic anal atresia: sequencing of candidate genes, array-based molecular karyotyping, and review of the literature. European Journal of Pediatrics, 2011, 170, 741-746.	2.7	16
130	De novo duplication of 18p11.21â€“18q12.1 in a female with anorectal malformation. American Journal of Medical Genetics, Part A, 2011, 155, 445-449.	1.2	20
131	Bias in patient series with VACTERL association. American Journal of Medical Genetics, Part A, 2011, 155, 2039-2041.	1.2	9
132	Genome-wide expression profiling of urinary bladder implicates desmosomal and cytoskeletal dysregulation in the bladder exstrophy-epispadias complex. International Journal of Molecular Medicine, 2011, 27, 755-65.	4.0	19
133	p63 (TP73L) a key player in embryonic urogential development with significant dysregulation in human bladder exstrophy tissue. International Journal of Molecular Medicine, 2010, 26, 861-7.	4.0	19
134	Research perspectives in the etiology of congenital anorectal malformations using data of the International Consortium on Anorectal Malformations: evidence for risk factors across different populations. Pediatric Surgery International, 2010, 26, 1093-1099.	1.4	58
135	Embryonic expression of the cysteine rich protein 61 (CYR61) gene: A candidate for the development of human epispadias. Birth Defects Research Part A: Clinical and Molecular Teratology, 2010, 88, 546-550.	1.6	7
136	Evidence for linkage of the bladder exstrophyâ€epispadias complex on chromosome 4q31.21â€22 and 19q13.31â€41 from a consanguineous iranian family. Birth Defects Research Part A: Clinical and Molecular Teratology, 2010, 88, 757-761.	1.6	7
137	Genome-wide association study identifies two susceptibility loci for nonsyndromic cleft lip with or without cleft palate. Nature Genetics, 2010, 42, 24-26.	21.4	379
138	Microduplications at 22q11.21 are associated with non-syndromic classic bladder exstrophy. European Journal of Medical Genetics, 2010, 53, 55-60.	1.3	45
139	Genomeâ€wide linkage scan of nonsyndromic orofacial clefting in 91 families of central European origin. American Journal of Medical Genetics, Part A, 2009, 149A, 2680-2694.	1.2	38
140	Genomeâ€wide linkage scan for bladder exstrophyâ€epispadias complex. Birth Defects Research Part A: Clinical and Molecular Teratology, 2009, 85, 174-178.	1.6	20
141	Bladder exstrophyâ€epispadias complex. Birth Defects Research Part A: Clinical and Molecular Teratology, 2009, 85, 509-522.	1.6	53
142	Possible association of Down syndrome and exstrophyâ€“epispadias complex: report of two new cases and review of the literature. European Journal of Pediatrics, 2009, 168, 881-883.	2.7	5
143	Key susceptibility locus for nonsyndromic cleft lip with or without cleft palate on chromosome 8q24. Nature Genetics, 2009, 41, 473-477.	21.4	415
144	Transforming growth factor-beta receptor type 1 (TGFBR1) is not associated with non-syndromic cleft lip with or without cleft palate in patients of Central European descent. International Journal of Pediatric Otorhinolaryngology, 2009, 73, 1334-1338.	1.0	3

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145	The Exstrophy-epispadias complex. Orphanet Journal of Rare Diseases, 2009, 4, 23.	2.7	198
146	TGFB3 displays parent-of-origin effects among central Europeans with nonsyndromic cleft lip and palate. Journal of Human Genetics, 2008, 53, 656-661.	2.3	34
147	Investigation of FGF10 as a candidate gene in patients with anorectal malformations and exstrophy of the cloaca. Pediatric Surgery International, 2008, 24, 893-897.	1.4	22
148	Family-Based Association Study of the MTHFR Polymorphism C677T in Patients with Nonsyndromic Cleft Lip and Palate from Central Europe. Cleft Palate-Craniofacial Journal, 2008, 45, 267-271.	0.9	19
149	A family-based association study in Central Europeans: No evidence for the cystathionine beta-synthase c.844ins68 gene variant as a risk factor for non-syndromic cleft lip and palate. American Journal of Medical Genetics, Part A, 2007, 143A, 205-207.	1.2	6
150	Concordance analyses of twins with bladder exstrophyâ€“epispadias complex suggest genetic etiology. American Journal of Medical Genetics, Part A, 2007, 143A, 2751-2756.	1.2	53
151	Genome-wide analysis for micro-aberrations in familial exstrophy of the bladder using array-based comparative genomic hybridization. BJU International, 2007, 100, 646-650.	2.5	12
152	Bladder exstrophyâ€“epispadias complex: Investigation of suppressor of variegation, enhancer of zeste and Trithorax (SET) as a candidate gene in a large cohort of patients. Scandinavian Journal of Urology and Nephrology, 2006, 40, 221-224.	1.4	16
153	Gender-associated differences in the psychosocial and developmental outcome in patients affected with the bladder exstrophy-epispadias complex. BJU International, 2006, 97, 349-353.	2.5	45
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