

## **CURE-Net publication list**

### **ISL1 is a major susceptibility gene for classic bladder exstrophy and a regulator of urinary tract development**

Sci Rep. 2017 Feb 8;7:42170. doi: 10.1038/srep42170

Zhang R, Knapp M, Suzuki K, Kajioka D, Schmidt JM, Winkler J, Yilmaz Ö, Pleschka M, Cao J, Kockum CC, Barker G, Holmdahl G, Beaman G, Keene D, Woolf AS, Cervellione RM, Cheng W, Wilkins S, Gearhart JP, Sirchia F, Di Grazia M, Ebert AK, Rösch W, Ellinger J, Jenetzky E, Zwink N, Feitz WF, Marcelis C, Schumacher J, Martínón-Torres F, Hibberd ML, Khor CC, Heilmann-Heimbach S, Barth S, Boyadjiev SA, Brusco A, Ludwig M, Newman W, Nordenskjöld A, Yamada G, Odermatt B, Reutter H

### **Sexual function in adult patients with classic bladder exstrophy: A multicenter study**

J Pediatr Urol. 2015 Jun;11(3):125.e1-6. doi: 10.1016/j.jpurol.2015.02.001. Epub 2015 Mar 12.

Park W, Zwink N, Ebert A-K, Rösch W-H, Schmiedeke E, Stein R, Schmidt D, Noeker M, Jenetzky E, Reutter H.

### **Genome-wide association study and meta-analysis identify ISL1 as genome-wide significant susceptibility gene for bladder exstrophy**

PLoS Genet. 2015 Mar 12;11(3):e1005024. doi: 10.1371/journal.pgen.1005024. eCollection 2015

Draaken M, Knapp M, Pennimpede T, Ebert AK, Rösch W, Stein R, Utsch B, Hirsch K, Boemers TM, Mangold E, Heilmann S, Ludwig KU, Jenetzky E, Zwink N, Moebus S, Herrmann B, Mattheisen M, Nöthen M, Ludwig M.

### **European consensus meeting of ARM-Net members concerning diagnosis and early management of newborns with anorectal malformations**

Tech. Coloproctol. 2015 Mar;19(3):181-5. doi: 10.1007/s10151-015-1267-8. Epub 2015 Jan 22

Van der Steeg HJ, Schmiedeke E, Bagolan P, Broens P, Demirogullari B, Garcia-Vazquez A, Grasshoff-Derr S, Lacher M, Leva E, Makedonsky I, Sloots CE, Schwarzer N, Aminoff D, Schipper M, Jenetzky E, van Rooij IA, Giuliani S, Crétolle C, Holland Cunz S, Midrio P, de Blaauw I.

### **Genome-wide mapping of copy number variations in patients with both anorectal malformations and central nervous system abnormalities**

Birth Defects Res A Clin Mol Teratol 2014; [Epub ahead of print]

Dworschak GC, Draaken M, Hilger AC, Schramm C, Bartels E, Schmiedeke E, Grasshoff-Derr S, Märzheuser S, Holland-Cunz S, Lacher M, Jenetzky E, Zwink N, Schmidt D, Nöthen MM, Ludwig M, Reutter H.

### **Heterozygous FGF8 mutations in patients presenting cryptorchidism and multiple VATER/VACTERL features without limb anomalies**

Birth Defects Res A Clin Mol Teratol 2014; [Epub ahead of print]

Zeidler C, Woelfle J, Draaken M, Mughal SS, Große G, Hilger AC, Dworschak GC, Boemers TM, Jenetzky E, Zwink N, Lacher M, Schmidt D, Schmiedeke E, Grasshoff-Derr S, Märzheuser S, Holland-Cunz S, Schäfer M, Bartels E, Keppler K, Palta M, Leonhardt J, Kujath C, Reißmann A, Nöthen MM, Reutter H, Ludwig M.

### **Genome-wide association study and mouse expression data identify a highly conserved 32kb intergenic region between WNT3 and WNT9b as possible susceptibility locus for isolated classic exstrophy of the bladder**

Hum Mol Genet 2014; [Epub ahead of print]

Reutter H, Draaken M, Pennimpede T, Wittler L, Brockschmidt FF, Ebert AK, Bartels E, Rösch W, Boemers TM, Hirsch K, Schmiedeke E, Meesters C, Becker T, Stein R, Utsch B, Mangold E, Nordenskjöld A, Barker G, Kockum CC, Zwink N, Holmdahl G, Läckgren G, Jenetzky E, Feitz WF, Marcelis C, Wijers CH, van Rooij IA, Gearhart JP, Herrmann BG, Ludwig M, Boyadjiev SA, Nöthen MM, Mattheisen M.

**No major role for periconceptual folic acid use and its interaction with the MTHFR C677T polymorphism in the etiology of congenital anorectal malformations**

Birth Defects Res A Clin Mol Teratol 2014; 100(6):483-92

Wijers CH, de Blaauw I, Zwink N, Draaken M, van der Zanden LF, Brunner HG, Brooks AS, Hofstra RM, Sloots CE, Broens PM, Wijnen MH, Ludwig M, Jenetzky E, Reutter H, Marcelis CL, Roeleveld N, van Rooij IA

**Classic bladder exstrophy: Frequent 22q11.21 duplications and definition of a 414 kb phenocritical region**

Birth Defects Res A Clin Mol Teratol 2014; 100(6):512-7. 23.

Draaken M, Baudisch F, Timmermann B, Kuhl H, Kerick M, Proske J, Wittler L, Pennimpede T, Ebert AK, Rösch W, Stein R, Bartels E, von Lowtzow C, Boemers TM, Herms S, Gearhart JP, Lakshmanan Y, Kockum CC, Holmdahl G, Läckgren G, Nordenskjöld A, Boyadjiev SA, Herrmann BG, Nöthen MM, Ludwig M, Reutter H.

**Second study on the recurrence risk of isolated esophageal atresia with or without trachea-esophageal fistula among first-degree relatives: No evidence for increased risk of recurrence of EA/TEF or for malformations of the VATER/VACTERL association spectrum**

Birth Defects Res A Clin Mol Teratol 2013; 97(12): 786-91.

Choinitzki V, Zwink N, Bartels E, Baudisch F, Boemers TM, Hölscher A, Turial S, Bachour H, Heydweiller A, Kurz R, Bartmann P, Pauly M, Brokmeier U, Leutner A, Nöthen MM, Schumacher J, Jenetzky E, Reutter H.

**First results of a European multi-center registry of patients with anorectal malformations**

J Pediatr Surg 2013; 48(12): 2530-5.

De Blaauw I, Wijers CH, Schmiedeke E, Holland-Cunz S, Gamba P, Marcelis CL, Reutter H, Aminoff D, Schipper M, Schwarzer N, Grasshoff-Derr S, Midrio P, Jenetzky E, van Rooij IA.

**First results of a European multi-center registry of patients with anorectal malformations**

J Pediatr Surg 2013; 48(12): 2530-5.

De Blaauw I, Wijers CH, Schmiedeke E, Holland-Cunz S, Gamba P, Marcelis CL, Reutter H, Aminoff D, Schipper M, Schwarzer N, Grasshoff-Derr S, Midrio P, Jenetzky E, van Rooij IA.

**Whole-exome resequencing reveals recessive mutations in TRAP1 in individuals with CAKUT and VACTERL association**

Kidney Int 2014; 85(6): 1310-7

Saisawat P, Kohl S, Hilger AC, Hwang DY, Yung Gee H, Dworschak GC, Tasic V, Pennimpede T, Natarajan S, Sperry E, Matassa DS, Stajić N, Bogdanovic R, de Blaauw I, Marcelis CL, Wijers CH, Bartels E, Schmiedeke E, Schmidt D, Märzheuser S, Grasshoff-Derr S, Holland-Cunz S, Ludwig M, Nöthen MM, Draaken M, Brosens E, Heij H, Tibboel D, Herrmann BG, Solomon BD, de Klein A, van Rooij IA, Esposito F, Reutter HM, Hildebrandt F.

**Candidate gene association study implicates p63 in the etiology of nonsyndromic bladder-exstrophy-epispadias complex**

Birth Defects Res A Clin Mol Teratol 2013; 97(12): 759-63

Qi L, Wang M, Yagnik G, Mattheisen M, Gearhart JP, Lakshmanan Y, Ebert AK, Rösch W, Ludwig M, Draaken M, Reutter H, Boyadjiev SA.

**De novo Deletions of Chromosome 13q in Two Patients with Mild Anorectal Malformations as Part of VATER/VACTERL and VATER/VACTERL-like Association and Analysis of EFNB2 in Patients with Anorectal Malformations**

Am J Med Genet A, in press

Dworschak GC, Draaken M, Marcelis C, de Blaauw I, Pfundt R, van Rooij IALM, Bartels E, Hilger A, Jenetzky E, Schmiedeke E, Grasshoff-Derr S, Schmidt D, Märzheuser S, Hosie S, Weih S, Holland-Cunz S, Palta M, Leonhardt J, Schäfer M, Kujath C, Rißmann A, Nöthen MM, Zwink N, Ludwig M, Reutter H.

### **CNV Analysis in Monozygotic Twin Pairs Discordant for Urorectal Malformations**

Twin Res Hum Genet. 2013 May 9:1-6. [Epub ahead of print] PMID: 23659922

Baudisch F, Draaken M, Bartels E, Schmiedeke E, Bagci S, Bartmann P, Nöthen MM, Ludwig M, Reutter H.

### **De novo microduplications at 1q41, 2q37.3, and 8q24.3 in patients with VATER/VACTERL association**

Eur J Hum Genet. 2013 Apr 3. doi: 10.1038/ejhg.2013.58. [Epub ahead of print] PMID: 23549274

Hilger A, Schramm C, Pennimpede T, Wittler L, Dworschak GC, Bartels E, Engels H, Zink AM, Degenhardt F, Müller AM, Schmiedeke E, Grasshoff-Derr S, Märzheuser S, Hosie S, Holland-Cunz S, Wijers CH, Marcelis CL, van Rooij IA, Hildebrandt F, Herrmann BG, Nöthen MM, Ludwig M, Reutter H, Draaken M.

### **Isolated bladder exstrophy associated with a de novo 0.9 Mb microduplication on chromosome 19p13.12.**

Draaken M, Mughal SS, Pennimpede T, Wolter S, Wittler L, Ebert AK, Rösch W, Stein R, Bartels E, Schmidt D, Boemers TM, Schmiedeke E, Hoffmann P, Moebus S, Herrmann BG, Nöthen MM, Reutter H, Ludwig M. Birth Defects Res A Clin Mol Teratol Jan 2013;

### **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.**

Draaken M, Prins W, Zeidler C, Hilger A, Mughal SS, Latus J, Boemers TM, Schmidt D, Schmiedeke E, Spsychalski N, Bartels E, Nöthen MM, Reutter H, Ludwig M. Int J Mol Med 2012; 30(6): 1459-64.

### **Clinical geneticists' views of VACTERL/VATER association.**

Solomon BD, Bear KA, Kimonis V, de Klein A, Scott DA, Shaw-Smith C, Tibboel D, Reutter H, Giampietro PF. Am J Med Genet A 2012; 158A(12): 3087-100.

### **Assisted reproductive techniques and the risk of the exstrophy-epispadias complex: a German case-control study.**

Zwink N, Jenetzky E, Hirsch K, Reifferscheid P, Schmiedeke E, Schmidt D, Reckin S, Obermayr F, Boemers TM, Stein R, Reutter H, Rösch WH, Brenner H, Ebert AK. J Urol. Nov 2012;

### **Practice of dilatation after surgical correction in anorectal malformations (ARM).**

Jenetzky E, Reckin S, Schmiedeke E, Schmidt D, Schwarzer N, Grasshoff-Derr S, Zwink N, Bartels E, Reißmann A, Leonhardt J, Weih S, Obermayr F, Räddecke J, Palta M, Kosch F, Götz G, Hofbauer A, Schäfer M, Reutter H, Holland-Cunz S, Märzheuser S. Pediatr Surg Int. 2012; 28(11): 1095-9.

### **Transition of adolescents with the exstrophy-epispadias complex to adult medicine: influence of long-term outcome results on management**

Ebert AK, Reutter H, Neissner C, Rösch W. Klin Padiatr 2012; 224(7): 455-60.

### **VATER/VACTERL association: identification of seven new twin pairs, a systematic review of the literature, and a classical twin analysis.**

Bartels E, Schulz AC, Mora NW, Pineda-Alvarez DE, Wijers CH, Marcelis CM, Stressig R, Ritgen J, Schmiedeke E, Mattheisen M, Draaken M, Hoffmann P, Hilger AC, Dworschak GC, Baudisch F, Ludwig M, Bagci S, Müller A, Gembruch U, Geipel A, Berg C, Bartmann P, Nöthen MM, van Rooij IA, Solomon BD, Reutter HM. Clin Dysmorphol 2012; 21(4): 191-5.

### **Testicular tumors in patients with exstrophy-epispadias complex.**

Ebert AK, Kliesch S, Neissner C, Reutter H, Rösch WH. J Urol 2012; 188(4): 1300-5.

### **Murine expression and mutation analyses of the prostate androgen-regulated mucin-like protein 1 (Parm1) gene, a candidate for human epispadias.**

Wittler L, Hilger A, Proske J, Pennimpede T, Draaken M, Ebert AK, Rösch W, Stein R, Nöthen MM, Reutter H, Ludwig M. Gene 2012; 506(2): 392-5.

**Assisted reproductive techniques and the risk of anorectal malformations: a German case-control study.**

Zwink N, Jenetzky E, Schmiedeke E, Schmidt D, Märzheuser S, Grasshoff-Derr S, Holland-Cunz S, Weih S, Hosie S, Reifferscheid P, Ameis H, Kujath C, Reißmann A, Obermayr F, Schwarzer N, Bartels E, Reutter H, Brenner H, CURE-Net Consortium. *Orphanet J Rare Dis.* 2012; 7: 65.

**Postoperative complications in adults with anorectal malformation: a need for transition. German Network for Congenital Uro-Rectal Malformations (CURE-Net).**

Schmidt D, Jenetzky E, Zwink N, Schmiedeke E, Maerzheuser S. *Pediatr Surg Int.* 2012; 28(8): 793-5.

**Sexual function in adults with anorectal malformation: psychosocial adaptation. German Network for Congenital Uro-Rectal Malformations (CURE-Net).**

Schmidt D, Winter S, Jenetzky E, Zwink N, Schmiedeke E, Maerzheuser S. *Pediatr Surg Int.* 2012; 28(8): 789-92.

**Unexpected results of a nationwide, treatment-independent assessment of fecal incontinence in patients with anorectal anomalies.**

Schmiedeke E, Zwink N, Schwarzer N, Bartels E, Schmidt D, Grasshoff-Derr S, Holland-Cunz S, Hosie S, Jablonka K, Maerzheuser S, Reutter H, Lorenz C, Jenetzky E. *Pediatr Surg Int.* 2012; 28(8): 825-30.

**Familial occurrence of the VATER/VACTERL association.**

Hilger A, Schramm C, Draaken M, Mughal SS, Dworschak G, Bartels E, Hoffmann P, Nöthen MM, Reutter H, Ludwig M. *Pediatr Surg Int* 2012; 28(7): 725-9.

**Inheritance of the VATER/VACTERL association.**

Bartels E, Jenetzky E, Solomon BD, Ludwig M, Schmiedeke E, Grasshoff-Derr S, Schmidt D, Märzheuser S, Hosie S, Weih S, Holland-Cunz S, Palta M, Leonhardt J, Schäfer M, Kujath C, Reißmann A, Nöthen MM, Reutter H, Zwink N. *Pediatr Surg Int.* 2012; 28(7): 681-5.

**Phenotype Severity in the Bladder Exstrophy-Epispadias Complex: Analysis of Genetic and Nongenetic Contributing Factors in 441 Families from North America and Europe.**

Reutter H, Boyadjiev SA, Gambhir L, Ebert AK, Rösch WH, Stein R, Schröder A, Boemers TM, Bartels E, Vogt H, Utsch B, Müller M, Detlefsen B, Zwink N, Rogenhofer S, Gobet R, Beckers GM, Bökenkamp A, Kajbafzadeh AM, Jaureguizar E, Draaken M, Lakshmanan Y, Gearhart JP, Ludwig M, Nöthen MM, Jenetzky E. *J Pediatr.* 2011; 159(5): 825-31.

**Pregnancy management in women within the bladder-exstrophy-epispadias complex (BEEC) after continent urinary diversion.**

Ebert AK, Falkert A, Hofstädter A, Reutter H, Rösch WH. *Arch Gynecol Obstet.* 2011; 284(4): 1043-6.

**German Network for Congenital Uro-Rectal Malformations: first evaluation and interpretation of postoperative urological complications in anorectal malformations.**

Maerzheuser S, Jenetzky E, Zwink N, Reutter H, Bartels E, Grasshoff-Derr S, Holland-Cunz S, Hosie S, Schmiedeke E, Schwarzer N, Spychalski N, Goetz G, Schmidt D. *Pediatr Surg Int.* 2011; 27(10): 1085-9.

**Medical predictors of psychological anxieties in VATER patients.**

Noeker M, Schmitz M, Schmiedeke E, Zwink N, Reutter H, Schmidt D, Jenetzky E. *Pediatr Surg Int.* 2011; 27(10): 1079-83.

**A successful treatment strategy in infants and adolescents with anorectal malformation and incontinence with combined hydrocolonic ultrasound and bowel management.**

Grasshoff-Derr S, Backhaus K, Hubert D, Meyer T. *Pediatr Surg Int.* 2011; 27(10): 1099-103.

**Bias in patient series with VACTERL association.**

Jenetzky E, Wijers CHW, Marcelis CM, Zwink N, Reutter H, van Rooij IALM. *Am J Med Genet A. Am J Med Genet A.* 2011;155(8): 2039-41.

**De novo duplication of 18p11.21-18q12.1 in a female with anorectal malformation.**

Schramm C, Draaken M, Bartels E, Boemers TM, Schmiedeke E, Grasshoff-Derr S, Märzheuser S, Hosie S, Holland-Cunz S, Baudisch F, Priebe L, Hoffmann P, Zink AM, Engels H, Brockschmidt FF, Aretz S, Nöthen MM, Ludwig M, Reutter H. *Am J Med Genet A*. 2011; 155(2): 445-9.

**De novo Partial Trisomy 18p and Partial Monosomy 18q in a Patient with Anorectal Malformation.**

Bartels E, Draaken M, Kazmierczak B, Spranger S, Schramm C, Baudisch F, Nöthen MM, Schmiedeke E, Ludwig M, Reutter H. *Cytogenet Genome Res*. 2011; 134(3): 243-8.

**Parental risk factors and anorectal malformations: systematic review and meta-analysis.**

Zwink N, Jenetzky E, Brenner H. *Orphanet Journal of Rare Diseases* 2011; 6: 25.

**Genome-wide expression profiling of urinary bladder implicates desmosomal and cytoskeletal dysregulation in the bladder exstrophy-epispadias complex.**

Qi L, Chen K, Hur DJ, Yagnik G, Lakshmanan Y, Kotch LE, Ashrafi GH, Martinez-Murillo F, Kowalski J, Naydenov C, Wittler L, Gearhart JP, Draaken M, Reutter H, Ludwig M, Boyadjiev SA. *Int J Mol Med*. 2011; 27(6): 755-65.

**Mapping the NPHP-JBTS-MKS Protein Network Reveals Ciliopathy Disease Genes and Pathways.**

Sang L, Miller JJ, Corbit KC, Giles RH, Brauer MJ, Otto EA, Baye LM, Wen X, Scales SJ, Kwong M, Huntzicker EG, Sfakianos MK, Sandoval W, Bazan JF, Kulkarni P, Garcia-Gonzalo FR, Seol AD, O'Toole JF, Held S, Reutter HM, Lane WS, Rafiq MA, Noor A, Ansar M, Devi AR, Sheffield VC, Slusarski DC, Vincent JB, Doherty DA, Hildebrandt F, Reiter JF, Jackson PK. *Cell*. 2011; 145(4): 513-28.