Appendix. Summary of the translational research cited in the article

Disease	Reference	Year	Category	Methodology	Molecules and findings
Incontinence/OAB					
Urofacial syndrone	Ochoa [15]	1987	Clinical Study	Clinical Study	N/A
	Wang [17]	1997	Genetic locus	Genome mapping	10q23-q24
	Daly [18]	2010	Genomic mutation	Genome mapping	HPSE2 (heparanase-2)
	Stuart [19]	2014	Bialleric mutation	Genetic sequencing	LRIG2
Sporadic OAB	Aoki [11]	2009	Urinary Eicosanoid	ELISA	Prostaglandin E2
	Oktar [9]	2013	Urinary protein	ELISA	Nerve growth factor
	Telli [10]	2015	Urinary protein	ELISA	Nerve growth factor
Nocturnal enuresis					
Familial enuresis	Eiberg [28]	1995	Genetic locus	Genome mapping	13q13-q14.3 (ENUR1)
	Amell [29]	1997	Genetic locus	Genome mapping	12q (ENUR2)
	Eiberg [30]	1998	Genetic locus	Genome mapping	22q11 (ENUR3)
	Eiberg [31]	2001	Genetic locus	Genome mapping	4p16.1(ENUR4)
	Loeys [34]	2002	Genetic locus	Genome mapping	22q11, 13q13-14, 12q
Soradic enuresis	Wei [35]	2010	Polymorophism	PCR-RFLP	5-hydroxytryptamine receptor 2A gene
	Balat [36]	2007	Polymorophism	PCR-RFLP	Nitric oxide synthase gene polymorphisms
Diabetus insipidus	Müller [40]	2002	Genetic mutation	Targeted sequencing	Aquaporin-2 gene
	Kanemitsu [41]	2002	Genetic mutation	Targeted sequencing	Arginine vasopressin-neurophysin II
	Shalev [38]	2004	Genetic mutation	Targeted sequencing	Aquaporin-2
	Robben [43]	2007	Genetic mutation	Targeted sequencing	*V2R* gene deficiency
	Marshall [42]	2013	Clinical Study	Clinical Study	*WFS1* gene, encoding wolframin
	Yamashita [37]	2016	Genetic mutation	Targeted sequencing	*V2R* gene deficiency in partial DI
Posterior urethral valve	Boghossian [49]	2016	DNA CNV	WGS	47 rare candidate CNVs in 32 cases
Neurogenic bladder	Nishikawa [8]	2015	Protein	Immunostaining	PTH-PTH related protein receptor 1
Exstrophy	Draaken [51]	2015	Genetic mutation	GWAS	ISL1
	von Lowtzow [52]	2016	DNA CNV	WGS	Rare CNVs identified
Cloaca	Harrison [50]	2014	DNA CNV	WGS	CNV in 41% of caese, 5 gain and 2 loss
MMHIS	Wangler [53]	2014	Genetic mutation	WGS	*ACTG2* gene missense variants
	Thorson [54]	2014	Genetic mutation	WGS	*ACTG2* gene mutation

OAB, overactive bladder; N/A, not applicable; ELISA, enzyme-linked immunosorbent assay; PCR-RFLP, polymerase chain reaction-restriction fragment length polymorphism; DNA CNV, DNA copy number variant; DI, diabetes insipidus; WGS, whole genome sequencing; GWAS, genome-wide association study; MMHIS, megacystis microcolon intestinal hypoperistalsis.