Bartter subtype	**Gene**	**OMIM**	**Related Protein**	**Inheritance**	**prevalence**	**Clinical phenotype**	**Biochemical characteristics**	**Reference**
Type I	*SLC12A1*	601678	NKCC2 Na-K_2Cl cotransporter	AR	Rare disease with unknown prevalence. More common BS type. Accounts for 24 % of antenatal BS cases*	Polyhydramnios, preterm delivery, profound postnatal salt wasting, polyuria, risk of acute kidney injury, failure to thrive, nephrocalcinosis	Classic hypokalemic metabolic alkalosis, hypercalciuria,	[6]
Type II	*KCNJ1*	241200	ROMK Potassium channel	AR	Rare disease with unknown prevalence. More common BS type. Accounts for 25 % of antenatal BS cases*	Polyhydramnios, preterm delivery, postnatal salt wasting, failure to thrive, nephrocalcinosis	As type 1, transient hyperkalemia and metabolic acidosis may occur initially	[7]

Type III	*CLCNKB*	607364	ClC-Kb Chloride channel	AR	Rare disease with unknown prevalence. Maybe the most common type of BS worldwide. Accounts for 19 % of antenatal BS cases *	Highly variable onset from antenatal manifestation to school age (GS like BS 3), Polyhydramnios, preterm delivery, polyuria, failure to thrive, nephrocalcinosis	Severe electrolyte disturbances, profound hypokalemia, hypochloremia, variable calcium excretion, hypomagnesemia,	[1, 8]
Type IV A	*BSND*	602522	Barttin Beta subunit of CLC-Kb and CLC-Ka chloride channels	AR	Rare disease with unknown prevalence. Less common BS type. Accounts for 8 % of antenatal BS cases*	Polyhydramnios, preterm delivery massive postnatal salt wasting, polyuria, failure to thrive, undifferentiated, hyperechogenic kidney	Severe electrolyte disturbances postnatally, variable calcium excretions, Hypomagnesemia,	[9, 10]

						parenchyma, early development of chronic renal failure, **hearing loss**		
Type IV B	*CLCNKA* + *CLCNKB*	613090	ClC-Ka + ClC-Kb	DR	ultrarare BS type	as type IVa	as type IVa	[11]
Type V	*MAGED2*	300971	MAGED2 No direct transport	XLR	Rare disease with unknown prevalence. Less common BS type. Accounts for 9 % of antenatal BS cases[a]	Polyhydramnios, preterm delivery, postnatal salt wasting, polyuria, most severe antenatal manifestation, but transient character	Transient hypokalemic metabolic alkalosis, hypercalciuria	[12, 13]

Gitelman	*SLC12A3*	263800	NCCT Na-Cl cotransporter	AR	Most common tubulopathy worldwide ca. 1:5000[b]	Variable manifestation, mostly at school age or even incidental diagnosis, fatigue, muscle weakness, tetany, muscle cramps, synkopy	Hypokalemic metabolic alkalosis, hypomagnesemia, hypocalciuria	[4, 14]

Table S1: Bartter syndrome subtypes and Gitelman syndrome classification: AR, autosomal recessive; DR, digenic recessive; XLR, X-linked recessive;

[a] Prevalence in patients with antenatal BS, reported by Legrand et al. [15]; [b] Estimated incidence of GS reported by Kondo et al. [5].

Patient	Exome	Linked-read	Targeted long-fragment enrichment	Long-range PCR	Long-read WGS (PacBio)
P1.1	Yes			Yes (not sequenced)	
P1.2	Yes	Yes	Yes	Yes	
P1.3	Yes			Yes	
P2				Yes	
P3				Yes	
P4				Yes	Yes
P5				Yes	
P6				No[a]	Yes
P7			Yes	Yes	
P8				Yes	
P9			Yes	Yes	
P10				No[a]	Yes
P11			Yes	No[a]	
P12				Yes	
P13				Yes	
P14				Yes	
P15.1				Yes	
P15.2				Yes	
P16[b]				No[a]	
P17				Yes	
P18				Yes	

P19				No[a]	
P20				Yes	
P21				Yes	
P22				No[a]	
P23				Yes	
P24				Yes	
P25				No[a]	
P26[b]				No[a]	
P27				Yes	
P28				Yes	
P29				Yes	
P30				Yes	

Table S2: Molecular genetic analyses performed on each patient. [a] Long-range PCR was performed but no PCR fragment could be generated. [b] degraded DNA, low DNA quality.

Structural variant	Transposition haplotype 2.2kb	Transposition haplotype 3.0kb	Blue	Red	Light blue	Green	Orange	Purple	Light green	Yellow
Breakpoint coordinates (hg19)	chr1:16,360,478_16,362,445del ins[chr1:16,383,736_16,385,874]	chr1:16,360,478_16,363,413del ins[chr1:16,383,736_16,386,677]	chr1:16,363,803 chr1:16,387,061	chr1:16,360,478 chr1:16,383,736	chr1:16,363,772 chr1:16,387,030	chr1:16,362,445 chr1:16,385,874	chr1:16,353,336 chr1:16,375,142	chr1:16,357,595 chr1:16,379,321	chr1:16,386,677 chr1:16,363,413	chr1:16,366,847 chr1:16,389,355
P1.1[b]	X		X							
P1.2	X		X							
P1.3	X		X							
P2		X	X							
P3		X	X							
P4						X				
P5		X			X					
P6								X		
P7	X			X						
P8	X			X						
P9[a] allele 1	X			X						
P9[a] allele 2		X			X					
P10	X									X
P11			X				X		X	
P12		X	X							
P13		X	X							
P14[a] allele 1	X			X						
P14[a] allele 2		X			X					
P15.1						X				
P15.2						X				
P17	X			X						
P18[a]		X			X					
P20	X			X						

P21		X	X							
P23		X			X					
P24		X	X							
P27		X	X							
P28		X	X							
P29		X	X							
P30	X			X						

Table S3: Breakpoint-region coordinates found in this study. For easier readability, structural variants with ambiguous breakpoints inside homologous regions are described in accordance with the HGVS Recommendations for Sequence Variant Nomenclature, whereby the breakpoint is arbitrarily assigned to the 3'-most position allowed by the sequence context (3' rule). Colors listed behind the breakpoint coordinates refer to the colors used in Figure 4. [a] structural variant in compound heterozygosity. [b] No long-read sequencing done, but short-read WES shows identical genotype as both affected brothers.