

Supplementary Table 1. Continuous and categorical variables investigated for the association of history of kidney stones

Stone History	Investigated variable
Age	Covariate
Glucose (plasma)	Covariate
Urea (plasma)	Covariate
eGFR (CKD_EPI)	Covariate
Uric acid (plasma)	Covariate
Cholesterol	Covariate
Tryglicerides	Covariate
Cholesterol HDL	Covariate
Cholesterol LDL	Covariate
Albumin (plasma)	Covariate
ACR (urine)	Covariate
Weight	Covariate
Height	Covariate
Abdominal circumference	Covariate
Gluteal circumference	Covariate
BMI	Covariate
BSA	Covariate
Insulin	Covariate
Na (plasma)	Covariate
K (plasma)	Covariate
Cl (plasma)	Covariate
Ca (plasma)	Covariate
Phosphate (plasma)	Covariate
hsCRP	Covariate
Cystatin_C (plasma)	Covariate

Creatinine (plasma)	Covariate
Systolic BP	Covariate
Diastolic BP	Covariate
CKD_stage	Covariate
Q: Hypertension	Covariate
Q: Drugs for hypertension	Covariate
Q: Previous/known renal diseases	Covariate
Q: Renal diseases in the family	Covariate
MS	Covariate
Gender	Covariate

Legend: ACR, albumin/creatinine ratio; BP, blood pressure; BSA, body surface area; hsCRP, high sensitivity C reactive protein; MS, Metabolic syndrome; Q: question on;

Supplementary Table 2. Variants of candidate genes associated with stone history

INCIPE 1

MARKER							SH			NSH		
CHR	POS	REF	ALT	RSCODE	CLNSIG	GENE	0/0	0/1	1/1	0/0	0/1	1/1
chr1	180634486	G	A	rs138879807	-	XPR1	124	7	0	750	12	0
chr1	180646152	G	A	rs3903981	-	XPR1	3	31	97	26	250	486
chr1	180669434	G	A	rs145177695	-	XPR1	125	6	0	691	71	0
chr1	180723190	C	T	rs12040223	-	XPR1	124	7	0	748	14	0
chr1	180742802	T	C	rs2331753	-	XPR1	3	33	95	26	249	487
chr1	180769656	G	A	rs114197278	-	XPR1	124	7	0	750	12	0
chr1	180777554	A	G	rs116112940	-	XPR1	125	6	0	686	76	0
chr1	180846345	A	G	rs72707405	-	XPR1	95	33	3	489	247	26
chr1	180851586	C	A	rs116688930	-	XPR1	125	6	0	687	75	0
chr1	180851588	A	T	rs114203991	-	XPR1	125	6	0	687	75	0
chr1	180866104	C	T	rs138447792	-	XPR1	124	7	0	749	13	0
chr1	180873358	A	C	rs75831517	-	XPR1	127	4	0	705	57	0
chr7	138714582	A	C	rs3080494	-	ATP6V0A4	89	39	3	584	165	13
chr7	138716422	T	C	rs6943456	-	ATP6V0A4	87	39	5	563	180	19
chr7	138724665	A	G	rs7808193	-	ATP6V0A4	86	40	5	563	180	19
chr7	138725596	T	C	rs34587733	-	ATP6V0A4	86	40	5	565	178	19

chr7	138726967	T	C	rs6973199	-	ATP6V0A4	89	37	5	579	165	18
chr7	138726981	G	C	rs34093718	-	ATP6V0A4	110	21	0	704	54	4
chr7	138730130	C	T	rs1809086	-	ATP6V0A4	43	59	29	289	380	93
chr7	138730334	C	T	rs896191	-	ATP6V0A4	8	38	85	64	317	381
chr7	138730386	G	A	rs896190	-	ATP6V0A4	8	38	85	65	325	372
chr7	138730482	G	A	rs1473390	-	ATP6V0A4	78	48	5	535	202	25
chr7	138730939	A	T	rs2353838	-	ATP6V0A4	6	33	92	45	289	428
chr7	138731410	G	A	rs72611547	-	ATP6V0A4	35	60	36	242	386	134
chr7	138732592	G	A	rs3778694	-	ATP6V0A4	33	62	36	240	386	136
chr7	138732973	A	G	rs3807154	Benign	ATP6V0A4	7	39	85	58	321	383
chr7	138735214	A	G	rs2122664	-	ATP6V0A4	5	28	98	24	257	481
chr7	138742075	T	C	rs10249244	-	ATP6V0A4	82	43	6	528	220	14
chr7	138745470	A	G	rs35974644	-	ATP6V0A4	109	22	0	700	59	3
chr7	138748254	A	C	rs4728445	-	ATP6V0A4	73	49	9	497	241	24
chr7	138748438	G	A	rs4732328	-	ATP6V0A4	73	49	9	497	241	24
chr7	138748761	A	C	rs7781292	-	ATP6V0A4	73	48	10	484	253	25
chr7	138748801	C	T	rs61392013	-	ATP6V0A4	116	14	1	707	55	0
chr7	138749740	T	C	rs366193	-	ATP6V0A4	17	56	58	131	367	264
chr7	138750001	T	C	rs367253	-	ATP6V0A4	52	57	22	348	329	85
chr7	138751070	A	C	rs451054	-	ATP6V0A4	17	57	57	133	370	259

chr7	138751122	A	G	rs390798	-	ATP6V0A4	17	57	57	132	371	259
chr7	138752135	T	C	rs427031	-	ATP6V0A4	11	60	60	107	357	298
chr7	138752241	G	T	rs441528	-	ATP6V0A4	17	57	57	133	372	257
chr7	138752389	C	G	rs28491851	-	ATP6V0A4	17	56	58	133	371	258
chr7	138753694	C	G	rs423184	-	ATP6V0A4	17	56	58	132	373	257
chr7	138753772	T	A	rs453407	-	ATP6V0A4	17	55	59	132	372	258
chr7	138754103	T	C	rs411467	-	ATP6V0A4	52	57	22	350	327	85
chr7	138756379	A	G	rs296910	-	ATP6V0A4	17	53	61	130	365	267
chr7	138757851	T	C	rs296909	-	ATP6V0A4	17	54	60	130	365	267
chr7	138766325	C	T	rs34665778	-	ATP6V0A4	112	19	0	708	52	2
chr7	138773192	G	A	rs35336496	-	ATP6V0A4	113	18	0	710	51	1
chr7	138783773	G	A	rs11974755	-	ATP6V0A4	118	13	0	636	116	10
chr8	42483325	C	T	rs112844178	-	SCL20A2	125	6	0	662	99	1
chr8	42487905	C	T	rs117673501	-	SCL20A2	124	7	0	744	18	0
chr8	42489432	C	T	rs117173054	-	SCL20A2	132	8	0	743	19	0
chr8	42489510	T	C	rs57699438	-	SCL20A2	123	8	0	741	21	0
chr8	42490887	CTT	C	rs146551698	-	SCL20A2	123	8	0	740	22	0
chr8	42503375	C	T	rs74857652	-	SCL20A2	130	1	0	720	39	3
chr12	4374636	C	T	rs10774226	-	FGF23	2	46	83	41	285	436
chr20	54161174	G	A	rs2585440	-	CYP24A1	119	11	1	719	43	0

chr20	54163591	ACC	A	rs148328441	-	CYP24A1	120	11	0	731	31	0
chr20	54164712	G	C	rs2762939	-	CYP24A1	67	55	9	456	269	37
chr20	54165160	C	T	rs76151159	-	CYP24A1	120	11	0	732	30	0
chr20	54165411	A	C	rs2762940	-	CYP24A1	67	55	9	455	270	37
chr20	54165489	T	C	rs76966109	-	CYP24A1	120	11	0	732	30	0
chr20	54165512	A	G	rs2585429	-	CYP24A1	39	60	32	286	361	115
chr20	54166666	A	C	rs78851953	-	CYP24A1	124	7	0	745	17	0
chr20	54167770	CAAAT	C	rs5841977	-	CYP24A1	61	56	14	421	291	50
chr20	54168073	T	C	rs2585431	-	CYP24A1	15	55	61	112	374	276
chr20	54170763	A	G	rs13038432	-	CYP24A1	120	11	0	633	126	3
chr20	54171755	C	A	rs36106327	-	CYP24A1	119	12	0	734	28	0
chr20	54172103	A	G	rs79276611	-	CYP24A1	119	12	0	730	32	0
chr20	54173157	G	A	rs35792925	-	CYP24A1	119	12	0	730	32	0

INCIPE 2

MARKER							SH			NSH		
CHR	POS	REF	ALT	RSCODE	CLNSIG	GENE	0/0	0/1	1/1	0/0	0/1	1/1
chr1	180631083	G	T	rs3819952	-	XPR1	208	36	1	1414	347	30
chr1	180631084	C	T	rs3819951	-	XPR1	208	36	1	1414	347	30
chr1	180706787	A	T	rs535665609	-	XPR1	237	8	0	1762	29	0

chr1	180848299	C	T	rs145804446	-	XPR1	238	7	0	1762	29	0
chr2	70957364	T	G	rs56237258	-	ATP6V1B1	144	87	14	1120	604	67
chr5	177386231	GGTCCCCAAGCTGCGCCAGGCT	G	rs876661296	Benign	SLC34A1	233	12	0	1738	53	0
chr8	42473720	C	G	rs140854316	-	SLC20A2	229	16	0	1721	68	2
chr9	137233633	T	C	rs28407527	Benign	SLC34A3	91	123	31	585	871	335
chr12	4376738	C	T	rs13312762	-	FGF23	228	17	0	1727	63	1
chr20	54162671	G	T	rs6091828	-	CYP24A1	218	27	0	1679	111	1
chr20	54163591	ACC	A	rs148328441	-	CYP24A1	227	18	0	1708	83	0
chr20	54164943	C	T	rs76194329	-	CYP24A1	219	26	0	1678	112	1
chr20	54165160	C	T	rs76151159	-	CYP24A1	227	18	0	1708	83	0
chr20	54165489	T	C	rs76966109	-	CYP24A1	227	18	0	1708	83	0
chr20	54168073	T	C	rs2585431	-	CYP24A1	27	111	107	253	851	687
chr20	54171755	C	A	rs36106327	-	CYP24A1	227	18	0	1710	81	0
chr20	54172103	A	G	rs79276611	-	CYP24A1	225	20	0	1709	80	2
chr20	54173157	G	A	rs35792925	-	CYP24A1	225	20	0	1707	82	2

Supplementary Table 3. Significant variants in INCIPE and already described in other papers

		NSH (n. 3386)		SH (n. 478)	
	Variants described by Halbritter et al. 2015 Braun et al. 2016 and observed in INCIPE	number of heterozygous subjects (%)	number of homozygous subjects (%)	number of heterozygous subjects (%)	number of homozygous subjects (%)
SLC34A1	chr5:177386231:GGTCCCAA GCTGCGCCAGGCT:G rs876661296	53 (1.6)	0	12 (2.5)	0
CYP24A1	chr20:54153445:C:T rs4811494	605 (17.9)	89 (2.6)	96 (20.1)	9 (1.9)

Jan Halbritter et al.: J Am Soc Nephrol 26: 543–551, 2015. doi: 10.1681/ASN.2014040388

Daniela Anne Braun et al.: Clin J Am Soc Nephrol 11: 664–672, 2016. doi: 10.2215/CJN.07540715

Supplementary Table 4. Pathogenetic variants of chromosome 20

MARKER							INCIPE1 SH			INCIPE1 NSH			INCIPE2 SH			INCIPE2 NSH			TOTAL SH			TOTAL NSH			TOTAL SH PERC			TOTAL NSH PERC		
CHR	POS	REF	ALT	GENE	RSCODE	CLNSIG	0/0	0/1	1/1	0/0	0/1	1/1	0/0	0/1	1/1	0/0	0/1	1/1	0/0	0/1	1/1	0/0	0/1	1/1	0/0	0/1	1/1	0/0	0/1	1/1
chr20	54157426	G	A	CYP24A1	NA	Pathogenic	131	0	0	762	0	0	245	0	0	1791	0	0	376	0	0	2553	0	0	1	0	0	1	0	0
chr20	54158135	C	T	CYP24A1	rs143934667	Pathogenic	131	0	0	762	0	0	245	0	0	1790	1	0	376	0	0	2552	1	0	1	0	0	0,9996083	0,0003917	0
chr20	54158136	G	A	CYP24A1	rs114368325	Pathogenic	129	2	0	755	7	0	244	1	0	1781	10	0	373	3	0	2536	17	0	0,99202128	0,00797872	0	0,99334117	0,00665883	0
chr20	54158967	C	G	CYP24A1	NA	Pathogenic	131	0	0	762	0	0	245	0	0	1791	0	0	376	0	0	2553	0	0	1	0	0	1	0	0
chr20	54159075	G	A	CYP24A1	rs777947329	Pathogenic/Likely_pathogenic	131	0	0	762	0	0	245	0	0	1791	0	0	376	0	0	2553	0	0	1	0	0	1	0	0
chr20	54159107	ATTAGACTG	A	CYP24A1	rs770055617	Pathogenic	131	0	0	762	0	0	245	0	0	1791	0	0	376	0	0	2553	0	0	1	0	0	1	0	0
chr20	54162743	C	T	CYP24A1	rs387907324	Pathogenic	131	0	0	762	0	0	245	0	0	1791	0	0	376	0	0	2553	0	0	1	0	0	1	0	0
chr20	54173045	CA	C	CYP24A1	NA	Pathogenic	131	0	0	762	0	0	245	0	0	1791	0	0	376	0	0	2553	0	0	1	0	0	1	0	0
chr20	54173517	CG	C	CYP24A1	rs774432244	Pathogenic/Likely_pathogenic	131	0	0	762	0	0	245	0	0	1791	0	0	376	0	0	2553	0	0	1	0	0	1	0	0
chr20	54173577	C	A	CYP24A1	rs781367354	Likely_pathogenic	131	0	0	762	0	0	245	0	0	1791	0	0	376	0	0	2553	0	0	1	0	0	1	0	0