

Sodium taurocholate cotransporting polypeptide (NTCP) deficiency: Identification of a novel *SLC10A1* mutation in two unrelated infants presenting with neonatal indirect hyperbilirubinemia and remarkable hypercholanemia

SUPPLEMENTARY MATERIALS

Supplementary Table 1: PCR primers and conditions for the sequencing analysis of UGT1A1 gene*

Targets	Primer sequences (5' to 3')	AT (°C)	Polymerase	Products (bp)
Promotor	Forward: AATGGATCCTGAGGTTCTGG Reverse: CTGGGTAGCCTCAAATTCCA	58	Taq	834
Exon 1	Forward: AGGAGCAAAGGCGCCATGGC Reverse: TGCCAAAGACAGACTCAAACC	58	Taq	1042
Exon 2	Forward: AACACGCATGCCTTTAATCATA Reverse: TGACAACAACCACAACAACAAA	56	Taq	416
Exon 3	Forward: GAAGTTGCCAGTCCTCAGAAG Reverse: TGTTGGCCATAATATTTTCAAGC	56	Taq	356
Exon 4	Forward: AACACTGAGTCTTTGGAGTGTTTTTC Reverse: TATTTGAAACAACGCTATTAATGCT	56	Taq	420
Exon 5	Forward: CAGGTTTCCTTTCCCAAGTTT Reverse: GAAGGCGTGTGTGTGTGAAC	60	Taq	1280

AT, annealing temperature; bp, base pairs.

*Based on the reference: Minucci A, Canu G, Gentile L, *et al.* Identification of a novel mutation in UDP- glucuronosyl transferase (UGT1A1) gene in a child with neonatal unconjugated hyperbilirubinemia. *Clinical Biochemistry*. 2013; 46:170–172.

Rodents and lagomorphs	Peptide	From	Amino acid sequence	To
Mouse	ENSMUSP00000093229	61	VIIAIVAQYGIPLSAFLLGKVFHLTSIEALA I	LICGCSPPGGNLSNLF 108
Ryukyu mouse	MGP_CAROLIEiJ_P0032680	61	VIIAIVAQYGIPLSAFLLGKVFHLTSIEALA I	LICGCSPPGGNLSNLF 108
Shrew mouse	MGP_PahariEiJ_P0082793	61	VIIAMVAQYGIPLSAFLLGKVFRLTNIIEALA I	LICGCSPPGGNLSNLF 108
Pika	ENSOPRP00000007592	61	LAIALVAQYGIPLTAFVLGKAFQMSKIEALA I	LICGCSPPGGNLSNLF 108
Rabbit	ENSOCUP00000008010	61	LAIALVAQYGIPLTAFVLGKVFRLTNIIEALA I	LICGCSPPGGNLSNLF 108
Brazilian guinea pig	ENSCAPP00000009872	61	LAIAMVAQYGIPLIAFLLGKVFSLTNIIEALA I	LICGCSPPGGNLSNLF 108
Chinese hamster CriGri	ENSCGRP00000012937	61	VVIAMLSQYGIPLAFAFVLGKAFHLKPIEALA I	LICGCSPPGGNLSNLF 108
Chinese hamster CHOK1GS	ENSCGRP0000001018980	61	VVIAMLSQYGIPLAFAFVLGKAFHLKPIEALA I	LICGCSPPGGNLSNLF 108
Damara mole rat	ENSFDAP00000016479	68	ITIGFLCQFGIMPLTGFVLSVAFGILPVQAVV V	IMGCCPPGGASSNVV 115
Degu	ENSODEP00000018011	61	LTIAMVAQYGIPLAFAFLLGKAFRLSNIIEALT L	LICGCSPPGGNLSNLF 108
Golden Hamster	ENSMAUP00000013002	61	VVIAMLSQYGIPLAFAFVLGKVFHLKPIEALA I	LICGCSPPGGNLSNLF 108
Guinea Pig	ENSCPOP00000005370	61	LAIAMVAQYGIPLTAFVLGKVFSLTNIIEALA I	LICGCSPPGGNLSNLF 108
Kangaroo rat	ENSODORP00000008282	69	IFVGFLLCQFGIMPLMGFLLSVAFHIIPIQAVV V	LIMGCCPPGGTASNVL 116
Long-tailed chinchilla	ENSCLAP00000006689	61	LAVAMVAQYGIPLTAFALGKMFRLNIEALA I	LICGCSPPGGNLSNLF 108
Naked mole-rat male	ENSHGLP00100024369	61	LATAMVAQYGIPLPFAFMLGKVFRLNIEALA I	LICGCSPPGGNLSNLF 108
Naked mole-rat female	ENSHGLP00000013534	61	LATAMVAQYGIPLPFAFMLGKVFRLNIEALA I	LICGCSPPGGNLSNLF 108
Northern American deer mouse	ENSPEMP00000022966	61	VVIAMLSQYGIPLTAFVLGKVFRLTNIIEALA I	LICGCSPPGGNLSNLF 108
Prairie vole	ENSMOCP00000016238	61	VVIAMLSQYGIPLAFAFVLGKVFRLNPIEALA I	LICGCSPPGGNLSNLF 108
Rat	ENSRNOP00000007825	61	VIVALVAQYGIPLAFAFLLGKIFHLSNIEALA I	LICGCSPPGGNLSNLF 108
Lesser Egyptian jerboa	ENSJJAP00000005005	61	LIIALVVSQYGIPLTAFVLGKVFRLTNIIEALA I	LICGCSPPGGNLSNLF 108
Squirrel	ENSSTOP00000003472	61	LAIAMVAQYGIPLTAFVLGKVFQLNIEALA I	LICGCSPPGGNLSNLF 108
Upper Galilee mountains blind mole rat	ENSNAP00000000640	61	LLIAMVAQYGIPLAFAFVLGKVFKLSNIEALA I	LICGCSPPGGNLSNLF 108

Other mammals	Peptide	From	Amino acid sequence	To
Cat	ENSFCAP00000008204	61	LAIALIAQYGIPLTAFALGKVFQLNIEALA I	LVCGCSPPGGTLSNLF 108
Dog	ENSCAFP00000024409	61	LVIALIAQYGIPLTAFLLGKVFRLNIEALA I	LVCGCSPPGGTLSNLF 108
Ferret	ENSMPUP00000007411	61	LAIALMAQYGIPLTAFLLGKVFRLNIEALA I	LVCGCSPPGGTLSNLF 108
Panda	ENSAMEP00000015380	61	LAIALIAQYGIPLTAFLLGKVFRLNIEALA I	LVCGCSPPGGTLSNLF 108
Alpaca	ENSVPAP00000005985	61	LAIALVAQYGIPLTAFSLGKLFRLNIEALA I	LICGCSPPGGNLSNLF 108
Cow	ENSBTAP00000002446	61	LAVALVAQYGIPLTAFGLGKFFQLNVEALA I	LICGCSPPGGNLSNLF 108
Dolphin	ENSTTRP00000005928	68	IAAGLLCQFGIMPLTAYLLVLSFSLKPVQAMA I	LIMGCCPPGGTISNLF 115
Horse	ENSECAP00000012659	61	LAIALVAQYGIPLTAFALGKVFQLNVEALA I	LVCGCSPPGGNLSNLF 108
Pig	ENSSSCP00000002511	61	LAIALVAQYGIPLTAFALGKLFRLNVEALA I	LICGCSPPGGNLSNLF 108
Sheep	ENSOARP00000022773	61	LAVALVAQYGIPLTAFGLGKFFQLNVEALA I	LICGCSPPGGNLSNLF 108
Armadillo	ENSNDOP00000004782	61	LVIALVAQYGIPLTAFVLGKVFRLNIEALA I	LICGCSPPGGTLSNLF 108
Elephant	ENSLAFP00000017315	61	LVALVAQYGIPLTAFGLGKLFRLNVEALA I	LICGCSPPGGNLSNLF 108
Hedgehog	ENSEEUP00000005963	68	IFVGFLLCQFGIMPLTGFLLSVAFGILPLQAVV V	LIMGCCPPGGTASNLF 115
Hyrax	ENSPCAP00000004134	61	LAIALVAQYGIPLTAFALGKLFRLNIEALA I	LICGCSPPGGNLSNLF 108
Lesser hedgehog tenrec	ENSETEP00000012656	61	LAIALVAQYGIPLTAFSLGKLFRLNIEALA I	LICGCSPPGGNLSNLF 108
Megabat	ENSPVAP00000009265	61	LAIALMARYGIPLTAFVLGKVFRLNIEALA I	LICGSSPPGGNLSNLF 108
Microbat	ENSMLUP00000010888	61	LAIAMVAQYGIPLTAFALGKLFRLNIEALA I	LICGCSPPGGNLSNLF 108
Shrew	ENSSARP00000000978	61	MIAMVAQYGIPLTAFSLGRIFKLNIEALA I	LVCGCSPPGGNLSNLF 108
Sloth	ENSCHOP00000004848	61	LAIALVAQYGIPLTAFALGKVFRLNIEALA I	LICGCSPPGGNLSNLF 108
Tree Shrew	ENSTBEP00000002346	61	LAIALLAQYGIPLTAFALGKVFPLNIEALA I	LVCGCSPPGGNLSNLF 108
Opossum	ENSMODP00000014214	69	IIIALVVSQYGVMLTAFSLGKIFHLNVEALA I	LICGCSPPGGTLSNLF 116
Platypus	ENSOANP00000017942	76	IIVGFLLCQFGIMPLTGFLLSLAFNVLPVQAVV I	IIMGCCPPGGTASNLF 123
Tasmanian devil	ENSSHAP00000003547	69	IAIALVVSQYGVMLTAFALGKIFQLNIEALA I	LICGCSPPGGTLSNLF 116
Wallaby	ENSMEUP00000007961	61	IAIALVVSQYGVMLTAFALGKIFRLNVEALA I	LICGCSPPGGTLSNLF 108

Other vertebrates	Peptide	From	Amino acid sequence	To
Anole lizard	ENSACAP00000005008	37	VMAAVSQFGIMMPLTAFALSIFRLAPMEALT V FIGCCPPGGTLSNIL	84
Chinese softshell turtle	ENSPSIP00000003539	76	ICVGFLCQFGIMPLTGFVLALLFNVLPAQAVA V LIMGCCPPGGTSSNIL	123
Chicken	ENSGALP00000054869	70	IFVGFLCQFGIMPLTAFLLSLAFDVHPIQAVV V MIMGCCPPGGTASNII	117
Duck	ENSAPLP00000014650	70	VAVAVMSQYGVMPPLTAFVLGKLFQLGPTESLA I LICGCCPPGGNLSNIF	117
Flycatcher	ENSFALP00000005168	62	VAVAIMAQYSIMPLTAFILGKLFQLGTSESLA I LICGCCPPGGNLSNIF	109
Turkey	ENSMGAP00000012582	58	VLLALLGQFVAMPLVAFLLALIFALDEVAAVA V LLCGCCPPGGNLSNLM	105
Zebra Finch	ENSTGUP00000011770	44	VAIAVLVQYSIMPLTAFILGKLFQLGTSESLA I LICGCCPPGGNLSNIF	91
Amazon molly	ENSPFOP00000017822	79	VAIAILSQYQIMPLTAFCLTKGFQLSEATAVV I LICGCCPPGGALSNIL	126
Cave fish	ENSAMP00000012264	75	VAIAVVAQYQIMPLSAFSLAKLLQLGPMETVA V LVCGCCPPGGNLSNIL	122
Cod	ENSGMOP00000017105	62	VAIAVVAQFGIMPLTAFCLATVFKLEQIEVLT V LICGCCPPGGNLSNIF	109
Coelacanth	ENSLACP00000008119	76	MAIAVVAQYQIMPLTAFALAKAFKLPTIESVT V LICGCCPPGGNLSNIF	123
Fugu	ENSTRUP00000037654	46	VAIALVAQFGIMPLTAFGLAKFLQMDAIKAVT V LICGCCPPGGTLSNIF	93
Medaka	ENSORLP00000021052	101	VAIALLAQFFIMPLSAFCLAKVLQMDPIKAVT V LICGCCPPGGTLSNIF	148
Platy fish	ENSXMAP00000007841	71	VAIAILSQYQIMPLTAFCLTKGFQVPEATAVV I LICGCCPPGGAMSNIL	117
Spotted gar	ENSLOCP00000015305	76	VGIAVIAQYGVMPITAFALAKMFQLGAEIAVT V LICGCCPPGGNLSNIF	123
Stickleback	ENSGACP00000010376	83	LIIAVVSQYVVMPLVAFCLVQAFQLTEIRAVV V LICGCCPPGGNLSNIL	130
Tetraodon	ENSTNIP00000020983	72	IVVGFLCQFGIMPFTAFALALAFQVI PVQAVV I IIMGCCPPGGTGSNII	119
Tilapia	ENSONIP00000007266	82	VAIAVVAQYQIMPLTAFCLAKLFKLAIEIAVV I LICGCCPPGGALSNIL	129
Zebrafish	ENSDDARP00000047825	72	VVIAVGAQYQIMPFTAFCLAKLFRLSPMESLS V LICGCCPPGGNLSNIF	119
Xenopus	ENSXETP00000059800	69	IGIALFSQFGIMPLAAFLAHVFLQNAIESVT V LICGCCPPGGVLSVVF	116
Lamprey	ENSPMAP00000005222	76	IFIAVLAQFGIMPLTAFALAKVFRLNPVEAVT V LICGCCPPGGNLSNVF	123

Other species	Peptide	From	Amino acid sequence	To
Caenorhabditis elegans	Y71G10AR.4 pep	219	PGIGLFAQFIIMPLLSYLIAYAIFMPRGLYSM A LGLFVTGCSPPGGAS	266
Ciona intestinalis	ENSCINP00000008688	45	IATAASCQFGIMPFISFLMAKIFGLDKVAAIA V LVTGCCPPGGNLSNLL	93
Ciona savignyi	ENSCSAVP00000001569	61	IALVAQFLIMPASAFGLTQAFQLDYAAIAVL I CGCCPPGGNLSNMLAY	108
Fruitfly	FBpp0074072	188	PVVGVSREVLMPAVGFLGRALWPDSPWLQLA L FYTALSPSGGLANVC	235

Supplementary Figure 1: Comparative alignment of the homologous peptides in rodents and lagomorphs, other mammals, other vertebrates as well as other species.