Title (en)
METHOD TO DIAGNOSE AND TREAT PATHOLOGICAL CONDITIONS RESULTING FROM DEFICIENT ION TRANSPORT

Title (de)
VERFAHREN ZUR DIAGNOSE UND BEHANDLUNG VON PATHOLOGISCHEN ZUSTÄNDEN IN ZUSAMMENHANG MIT IONENTRANSPORTDEFIZIENZEN

Title (fr)
PROCEDE POUR DIAGNOSTIQUER ET TRAITER DES TROUBLES PATHOLOGIQUES LIES A UNE INSUFFISANCE DU TRANSPORT D'IONS

Publication
EP 1030858 A1 20000830 (EN)

Application
EP 97954793 A 19971219

Priority
• US 9723553 W 19971219
• US 77805296 A 19961231

Abstract (en)
[origin: WO9829431A1] The present invention is based, in part, on the identification of the roles of the human thiazide-sensitive Na-Cl cotransporter, TSC; the human ATP-sensitive K$<+>$ channel, ROMK; and the human Na-K-2Cl cotransporter, NKCC2 in causing pathological condition associated with abnormal ion transport, particularly Bartter's Syndrome, Gitelman's Syndrome, hypokalaemic alkalosis, hypokalaemic alkalosis with hypercalciuria, kidney stones, high blood pressure, osteoporosis and sensitivity to diuretic-induced hyperkalaemia. The present invention specifically provides the amino acid sequence of several human wild-type and altered variants of the TSC, NKCC2 and ROMK proteins as well as the nucleotide sequence that encodes these variants that can be used in diagnosing ion transport disorders.

IPC 1-7
C07H 21/04; C12Q 1/68; C07K 14/435; C07K 16/00; G01N 33/53

IPC 8 full level
G01N 33/53 (2006.01); **C07K 14/435** (2006.01); **C07K 14/47** (2006.01); **C07K 14/705** (2006.01); **C07K 16/00** (2006.01); **C12N 1/15** (2006.01); **C12N 1/19** (2006.01); **C12N 1/21** (2006.01); **C12N 5/10** (2006.01); **C12N 15/09** (2006.01); **C12P 21/02** (2006.01); **C12Q 1/68** (2006.01); **G01N 33/68** (2006.01)

CPC (source: EP)
C07K 14/47 (2013.01); **C07K 14/705** (2013.01); **G01N 33/6893** (2013.01); G01N 2800/04 (2013.01); G01N 2800/34 (2013.01)

Citation (search report)
See references of WO 9829431A1

Designated contracting state (EPC)
AT BE CH DE DK ES FI FR GB GR IE IT LI LU MC NL PT SE

DOCDB simple family (publication)
WO 9829431 A1 19980709; AU 6013598 A 19980731; AU 746220 B2 20020418; CA 2274955 A1 19980709; EP 1030858 A1 20000830; JP 2001508291 A 20010626

DOCDB simple family (application)
US 9723553 W 19971219; AU 6013598 A 19971219; CA 2274955 A 19971219; EP 97954793 A 19971219; JP 53012398 A 19971219