

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

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Application

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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.

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