

Title (en)

HUMAN URIDINE DIPHOSPHATE GALACTOSE-4-EPIMERASE

Title (de)

MENSCHLICHE URIDINDIPHOSPHAT GALAKTOSE-4-EPIMERASE

Title (fr)

URIDINE DIPHOSPHATE GALACTOSE-4-EPIMERASE HUMAINE

Publication

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Application

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Priority

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Abstract (en)

[origin: WO9635778A1] A human UDP galactose-4-epimerase polypeptide and DNA (RNA) encoding such polypeptide and a procedure for producing such polypeptide by recombinant techniques is disclosed. Also disclosed are methods for utilizing such polypeptide for the treatment of galactosemia. Also disclosed are diagnostic assays for the detection of mutations in the nucleic acid sequencing encoding human UDP galactose-4-epimerase protein and for detecting an altered level of human UDP galactose-4-epimerase protein in a sample derived from a host.

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Citation (search report)

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- See references of WO 9635778A1

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