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[54] HUMAN GALACTOKINASE GENE 人類半乳糖激酶基因

[57]

[57] Abstract 撮錄

This invention relates to human galactokinase and the identification of galactokinase mutations, a missense and nonsense, as well as isolated nucleic acids encoding same, recombinant host cell transformed with DNA encoding such proteins and to uses of the expressed proteins and nucleic acid sequences in therapeutic and diagnostic applications.

此發明涉及人類半乳糖激酶與半乳糖激酶錯義和無義突變的鑒定,以及將其編碼的孤立核酸,用編碼這些蛋白質的DNA轉換的重組體宿主細胞,且涉及在治療和診斷應用中特殊蛋白質和核酸序列的用途。