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Cohen

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(54) **METHOD OF DIAGNOSING GALACTOSEMIA IN NEONATAL SCREENING**

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(58) **Field of Classification Search**

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See application file for complete search history.

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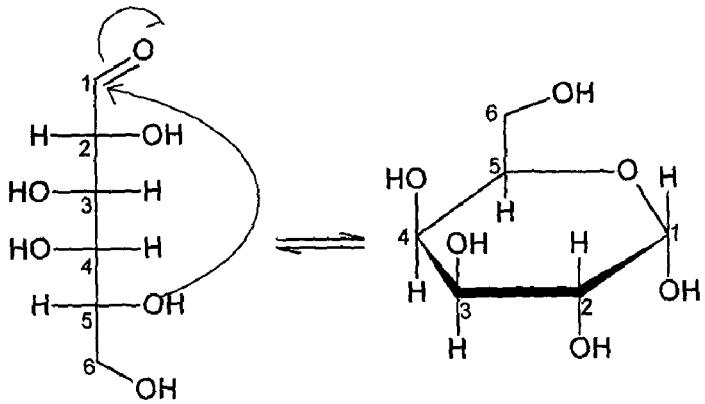
(57) **ABSTRACT**

A method of diagnosing galactosemia in blood samples from neonates by determining GAL-1-P concentrations before 5-7 days of life is disclosed. The removal of interfering compounds allows a more specific and therefore more accurate determination of GAL-IP levels in newborn screening for galactosemia using mass spectrometry. This is of major importance when investigating samples from children that have not yet achieved a steady state of GAL-IP (i.e. before 5-7 day of life).

20 Claims, 2 Drawing Sheets