

# Galactosemias

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Department of Oriental Pharmaceutical Sciences and Kyung Hee East-West- *Anal Biochem.* 2008 May 15;376(2):200-5. Epub 2008 Feb 29. ([DOI Direct Link](#))
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Dipartimento di Scienze Farmaceutiche, Universita di Pisa, Pisa, Italy.- *J Med Chem.* 2008 Jun 12;51(11):3182-93. Epub 2008 May 2. ([DOI Direct Link](#))
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Department of Human Genetics, Emory University School of Medicine,- *J Inherit Metab Dis.* 2008 Feb;31(1):97-107. Epub 2008 Jan 22. ([DOI Direct Link](#))

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Department of Human Genetics, Emory University School of Medicine, Emory- J Inherit Metab Dis. 2008 Feb;31(1):108-16. Epub 2008 Jan 14. ([DOI Direct Link](#))
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Centre de Reference des Maladies Hereditaires du Metabolisme et Inserm- J Hepatol. 2008 Mar;48(3):517-22. Epub 2007 Dec 31. ([DOI Direct Link](#))
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*Galactose metabolism occurs through an evolutionarily conserved pathway in...*28th May, 2008  
Leonard Miller School of Medicine, University of Miami, Miami, Florida,- Curr Protoc Hum Genet. 2008 Jan;Chapter 17:Unit 17.5. ([DOI Direct Link](#))
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Universitair Medisch Centrum Utrecht, divisie Kinderen, Huispost KC- Ned Tijdschr Geneesk. 2008 Mar 15;152(11):632-6.
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Department of Chemistry, McMaster University, Hamilton, ON, L8S 4M1,- Chem Commun (Camb). 2008 Jan 21;(3):338-40.
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*Galactose 1-phosphate uridylyltransferase deficiency causes the accumulation...*7th March, 2008  
- S Afr Med J. 2007 Dec;97(12):1231.
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Department of Reproduction and Gynecology, University Medical Center,- Reprod Sci. 2007 Dec;14(8):780-5. ([DOI Direct Link](#))

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