## PEDIATRIC GRAND ROUNDS Wednesday, April 6, 1960

## GALACTOSEMIA

because of bronchopneumonia and failure to thrive. The baby weighed 8 lbs. 1 oz. at birth and the neonatal course was uneventful except for mild jaundice. At 7 days of age, he had an episode of vomiting and his weight gain was always slow. At 6 months of age he weighed 8 lbs. 11 oz. and gained very little thereafter. For two months prior to admission he had frequent respiratory infections culminating in the bronchopneumonia which precipitated his admission. He was formula fed and his diet was confined to milk since it was stated that he vomited all other foods. The baby has never rolled over, cannot sit up and cannot crawl.

Family history shows that one of the father's relatives has diabetes. The mother is again pregnant.

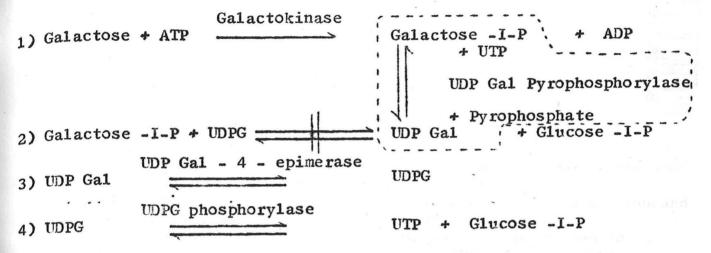
On examination he was a small baby, well below the third percentile for length and weight. The chest showed signs of a diffuse bronchopneumonia. The liver was palpable 2-3 fingers below the right costal margin. The eyes were normal. There was no jaundice.

Lab findings included a fasting blood sugar of 188 mg.% while on Similac, 64 mg.% while on nutramigen. Bilirubin 0.6 mg.%. Other chemistries were normal. The urine showed protein, casts and epithelial cells in addition to a reducing substance. An IVP was normal, but it was considered that the baby had meatal stenosis. The reducing substance in the urine was identified as galactose by paper chromatography.

The baby's hospital course is summarized in the chart. Following discharge, the baby has been doing much better and has been gaining 1 lb. per week on a milk free diet.

	Hospital Course	Doducina Cubatanca
Date	Diet	Reducing Substance in Urine
	Milk Nutramigen Restart Milk)	2% 0
, ,	Milk	2%
	Restart Nutramigen) Nutramigen	0

## Pathways of Galactose Metabolism



## References

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- 3. Kormsower, G.M. et al. A Clinical and Biochemical Study of Galactosemia Arch. Dis. Child. 31, 254, 1956
- 4. Holyel, A. et al. Galactosemia Am. J. Med. 22, 703, 1957
- 5. Isselbacher, K.J. et al. Congenital Galactosemia, a Single Enzymatic Block in Galactose Metabolism Science 123, 635, 1956
- 6. Anderson, E.P. et al. A Specific Enzymatic Assay for the Diagnosis of Congenital Galactosemia. J. Lab. Clin. Med. 50, 469, 1957.