



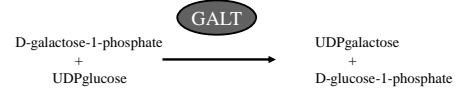
MALE ISSUES IN GALACTOSEMIA

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* No conflicts of interest to disclose

Galactosemia

Deficiency: galactose-1-phosphate-uridylyltransferase (GALT)



Gene: GALT on chromosome 9p13

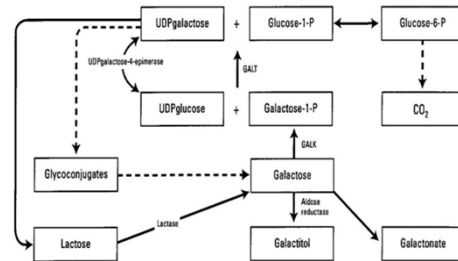
Frequency: 1/35,000 to 1/60,000 (1/16,476 in Ireland)

Inheritance: autosomal recessive

LONG-TERM CHRONIC COMPLICATIONS WITH NO CLEAR CAUSE

- Anxiety
- Depression
- Rare history of males who have been biological fathers
- Introverted or shy personalities (seems to affect males greater than females)

Galactose Metabolism



MALE FERTILITY

- Testicular size is normal
- Mean hormone levels were within normal range
- Sperm count tended to be in the low normal range
- Semen volume was reduced in 6 out of 8 men
- Two men tried to have children. One of these men and one other man fathered one child each.

Common GALT Mutations

- N314D
- Q188R
- S135L
- K285N
- L195P
- 5.2 kb deletion
- More than 230 GALT gene mutations have been detected

THANK YOU!

- We greatly thank the Galactosemia Foundation for supporting our work.