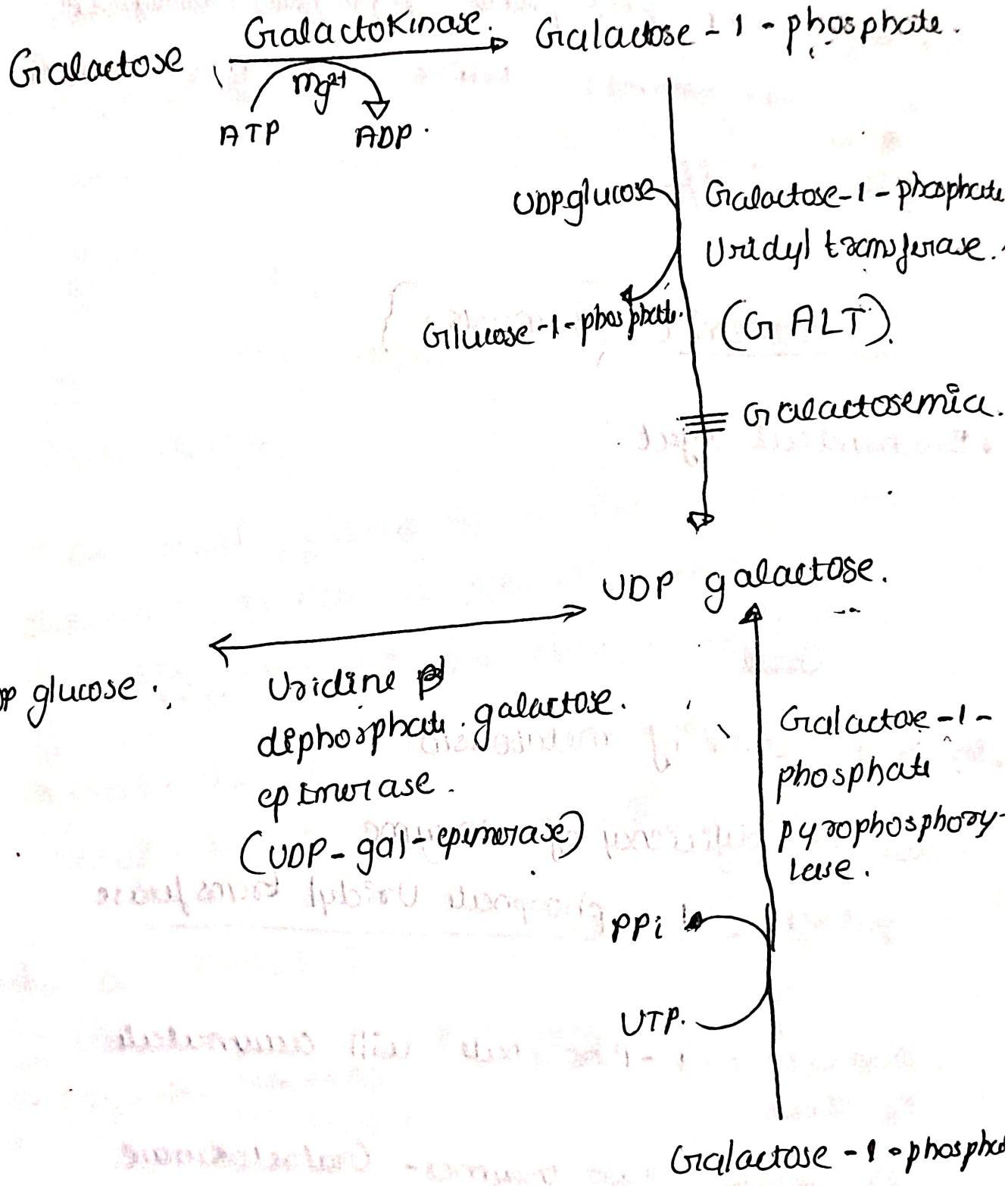
Galactose Metabolism

- Constituent of milk sugar (lactose)
- metabolized by liver.
- Galactose is used for,
  - synthesis of lactose, glycosaminoglycans, glycolipids, glycoproteins and cerebrosides.
- UDP galactose is the active donor of galactose during synthetic reactions.


Galactose
Galactokinase.
Galactose - 1 - phosphate.
Mg^{2+}
ATP
ADP.
UDPglucose
Galactose-1-phosphate
Uridyl transferase.
Glucose-1-phosphate.
(GALT).
Galactosemia.
UDP galactose.
UDP glucose.
Uridine
diphosphate galactose
epimerase.
(UDP-gal-epimerase)
Galactose-1-
phosphate
pyrophosphory-
lase.
PPi
UTP.
Galactose-1-phosphate

- Galactose-1-phosphate pyrophosphorylase in liver becomes active only after 4 or 5 years of life.

Galactosemia. {8 marks}

- Biochemical defect.

~~Gal~~

- In born error of metabolism.
- due to deficiency of enzyme galactose-1-phosphate uridyl transferase.

- Galactose-1-phosphate will accumulate in liver.
- It inhibit two enzymes - Galactokinase
  glycogen phosphorylase.

. Result in hypoglycemia, Hepatomegaly.

. Unconjugated bilirubin level increased due to defect in bilirubin conjugation in liver - jaundice.

. Severe mental retardation.

. Galactosuria.

- accumulated galactose is reduced to dulcitol, deposited in lens of eye and leads to congenital cataract due to it's osmotic effect.

. Galactose - 1 - phosphate accumulation in renal tubules leads to aminoaciduria.

. when a clinical qsn asked with

. Hepatomegaly
. congenital cataracts
} → . remember the disease.

Dignosis.

- prenatal diagnosis by amniocentesis
- Benedict's test.
- mucic acid test : specific test for galactose.

Treatment.

- Give lactose free diet.
- special diet may be withdrawn after 4 years, when galactose-1-phosphate pyrophosphorylase becomes active.

Galactokinase deficiency.

- Causes mild form of galactosemia.
  { Increased level of glactose in blood }
  - galactosuria.
- Congenital Cataracts is the main feature in this condition