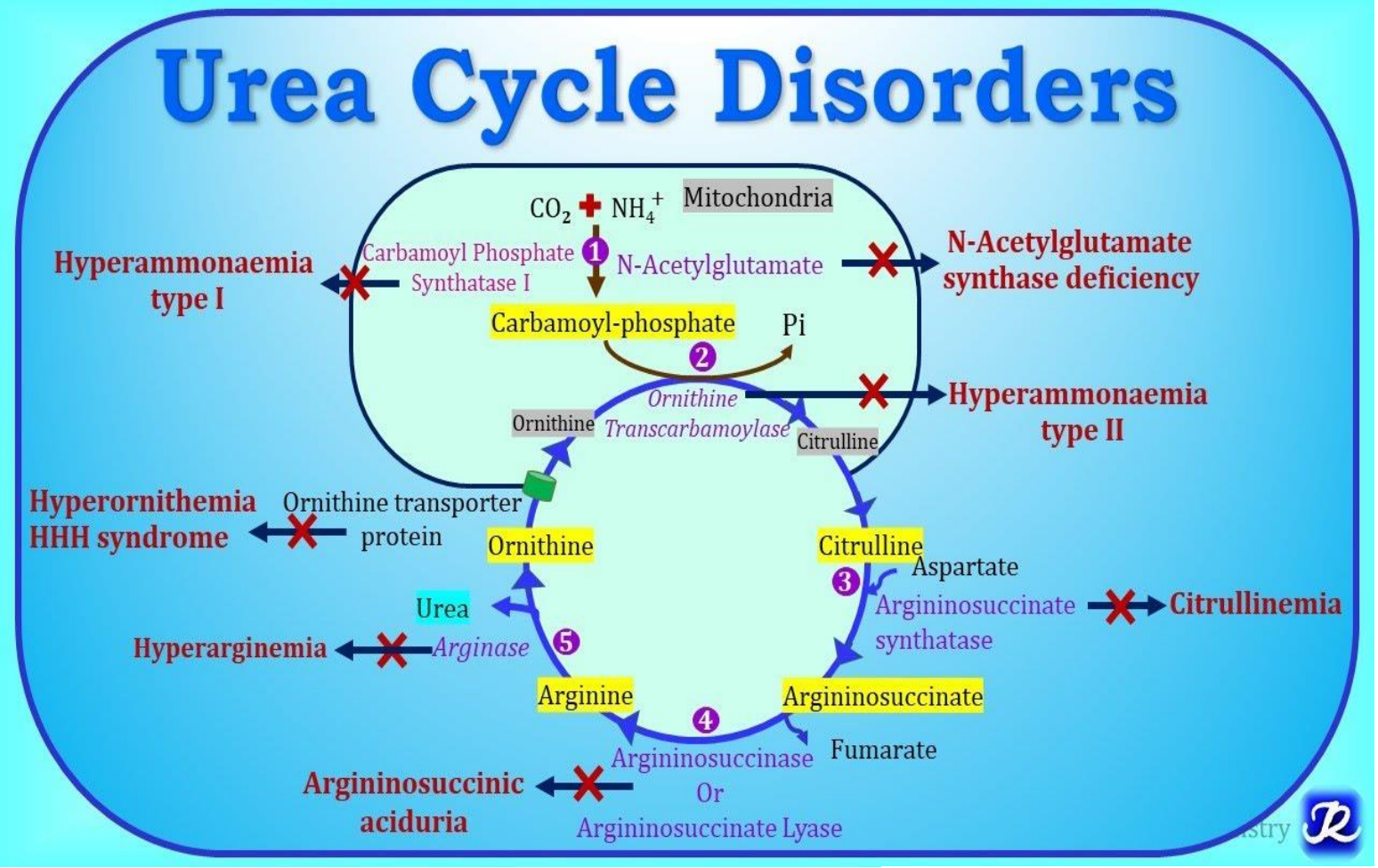

Defect	Enzyme involved
Hyperammonemia type I	Carbamoyl phosphate synthase I
Hyperammonemia type II	Ornithine transcarbamoylase
Citrullinemia	Arginosuccinate synthase
Arginosuccinic aciduria	Arginosuccinase
Hyperargininemia	Arginase

Urea cycle disorders		
Diseases	**Enzyme deficit**	**Features**
Hyperammonemia type I	CPS-I	Very high NH3 levels in blood. Autosomal recessive. Mental retardation.
Hyperammonemia type II	(OTC) Ornithine transcar-bamoy-lase	Ammonia level high in blood. Orotic aciduria due to channelling of carbamoyl phosphate into Pyrimidine synthesis. X-linked.
Hyperornithinemia	Defective ornithine trans-porter protein	Failure to import ornithine from cytoplasm to mitochondria. Defect in ORNT1 gene. Hyperornithinemia, hyperammonemia and homocitrullinuria is seen (HHH syndrome). Decreased urea in blood. Autosomal recessive condition.
Citrullinemia	Argininosuccinate synthe-tase	Autosomal recessive inheritance. High blood levels of ammonia and citrulline. Citrullinuria (1-2 g/day).
Argininosuccinic aciduria	Arginino-succinate lyase	Argininosuccinate in blood and urine. Friable brittle tufted hair (Trichorrhexis nodosa).
Hyperargininemia	Arginase	Arginine increased in blood and CSF.

Deficiency	Disorder	Clinical Feature
N-Acetylglutamate synthase	Hyperammonemia that may be accompanied by high plasma concentrations of alanine and glutamine	Lethargy; persistent vomiting; poor feeding; hyperventilation; enlarged liver; seizures
Carbamoyl phosphate synthetase	Hyperammonemia; citrullinemia; respiratory alkalosis	Lethargy; coma; seizures; vomiting; poor feeding; hyperventilation; hepatomegaly
Ornithine transcarbamylase	Hyperammonemia; respiratory alkalosis; elevated orotic acid in urine	Seizures; vomiting; poor feeding; hyperventilation; hepatomegaly
Arginosuccinate synthetase	Citrullinemia	Lethargy; coma; seizures; vomiting; poor feeding; hepatomegaly
Arginosuccinate lyase	Elevated arginosuccinic acid in urine	Lethargy; seizures; vomiting; poor feeding; hyperventilation; hepatomegaly
Arginase	Markedly elevated plasma arginine, lactate, and CSF glutamine, and modestly elevated blood ammonia	Delayed development; protein intolerance; spasticity; loss of muscle control; seizures; irritability

CSF indicates cerebrospinal fluid.