

Hyperammonemia

Age of onset of symptoms

<24 hr

>24 hr

Acidosis and/or ketosis

Preterm

Term

Present

Absent

Transient
hyperammonemia
of the newborn

Organic acid or
fatty acid oxidation
defect

Urea cycle
defect

Plasma citrulline

Absent-trace

100-300 μM

>1000 μM

Urine orotate

(+) plasma ASA
& anhydrides

Low

High

CPS
deficiency

OTC
deficiency

AL
deficiency

AS
deficiency