# 4. Citrullinemia: Clinical and Biochemical profile of Indian patients.

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**Objective:-** To determine clinical-biochemical profile of Indian children with Citrullinemia.

**Subjects and method:** We selected 316 critically ill babies admitted in NICUs with clinical suspicion of IMD. There were 7 babies with Citrullinemia (2.21%).

**Results: -** Of these 7 babies, 4 were males and 3 females. Two children were born out of 3rd degree consanguineous marriage (28.57%). The earliest presentation was 2nd day of life, latest 6th day. Of these 6 babies expired (85.71%). The mean values for biochemical parameters were: Ammonia 424 umol/L, Orotic Acid 763 umol/mmol Cr., Pl. Citrulline 1014 umol/L, Pl. Arginine 107.4 umol/L, Pl. Glutamine 958.3 umol/L and Pl. Ornithine 74.83 umol/L. All had normal Carnitine / Acyl Carnitine profile and urine GC-MS for Organic Acids. High Ur Orotic Acid picked up all cases in the very early stage. All except one were treated with sodium benzoate [SB] and one child received peritoneal dialysis in addition to SB. The only surviving child received Arginine, SB and special diet UrC1 from ComidaMed (Germany). She has excellent weight gain and is thriving well. Motor and mental milestones are normal. Though her Citrulline is always above 500umol/L, her ammonia is rarely above 60 umol/L and Ur. Orotic acid is always < 11umol/mmol Cr. All the families had lost atleast one child with similar disorder.

**Conclusion: -** All sick newborns should be screened for Citrullinemia as early treatment with SB, Arginine and proper diet may lead to good recovery. Urine Orotic acid screening is very helpful in early diagnosis.