

# Citrullinemia: Clinical and Biochemical profile of Indian Patients

Jalan A.<sup>1</sup>, Muehl A.<sup>2</sup>, Item Chike<sup>2</sup>, Bodamer O<sup>2</sup>, Shinde R.<sup>1</sup>, Telawane N.<sup>1</sup>,

1- *NIRMAN, Navi Mumbai Institute of Research in Mental and Neurological Handicap, India.* 2- *Biochemical Genetics and Newborn Screening Laboratories, University Children's Hospital, Vienna, Austria*

**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 3<sup>rd</sup> degree consanguineous marriage (28.57%). The earliest presentation was 2<sup>nd</sup> 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.

Citrullinemia	Marriage	Community	Past History	Outcome	Age	Sex
Name			Family History			
B/O Ch	Non Cons.	Hindu	1 Male Exp.	Expired	4 D	Male
B/O Sh	3rd Cons.	Muslim	3 Males Exp.	Expired	5 D	Male
B/O SC	Non Cons.	Hindu	1 Female Exp.	Expired	4 D	Female
B/O BS	3rd Cons.	Hindu	1 Male Exp.	Expired	2 D	Male
B/O SK	Non Cons.	Hindu	1 Female Exp.	Good	3 D	Female
B/O Ga	Non Cons.	Hindu	1 Male Exp.	Expired	6 D	Male
B/O KM	Non Cons.	Hindu	1 Male Exp.	Expired	3 D	Female

Citrullinemia	Ammonia	Orotic Acid	Ur. Creat	Ratio	Citrulline	Arginine	Glutamine	Ornithine	GC/MS of urine
Name	umol/L	umol/L	mmol/L	um/mm	uM/L	uM/L	uM/L	uM/L	
B/O Ch	455.00	NA	NA	NA	800.00	NA	NA	NA	Normal
B/O Sh	380.00	NA	NA	NA	2,469.00	20.00	3,239.00	107.00	Normal
B/O SC	620.00	114.04	2.65	43.00	920.00	13.65	473.50	36.00	Normal
B/O BS	1,000.00	7,371.00	2.51	2,935.00	1,700.00		518.50	81.00	Normal
B/O SK	840.00	2,185.00	9.61	227.39	2,200.00	214.00	1,209.00	40.00	Normal
B/O Ga	86.00	698.20	5.11	136.69	2,045.00		3,025.00	168.00	Normal
B/O KM		5,329.00	11.25	473.85	232.00	202.00	524.00	124.00	Normal
Average	424.0			763.0	1014.0	107.4	958.3	74.83	

**Conclusion:** - In India Citrullinemia appears to be relatively common UCD and 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. After completion of this study we had 3 more patients and now we are undertaking a project to identify molecular defects in our patients.

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