



IEM Incidence Rates

There are very few studies on IEM incidence rates in India. We have collated data from studies that have been presented in the Indian Journal of Pediatrics. One of the reasons that the incidence data is low is because no widespread studies have been done. We believe the rates are higher.

IEM incidence rates may vary according to populations but the cut off values for detection of the presence of IEMs are universal and does not depend on the population for the technology used by NeoGen Labs.

Incidence Rates - INDIA

Newborn Screening in India, A. Devi, S. Naushad, CDFD, IJP, Volume 71, February 2004. This paper presents the results from a study conducted in four major Government Maternity Hospitals in Hyderabad.

Metabolic Disorder	Screened	Confirmed	Incidence
Amino Acid	18,300	5	1:3600
Phenylketonuria	18,300	1	1:18300
Transient Tyrosinemia	18,300	2	1:9000
Hyperglycinemia	18,300	1	1:18300
Hyperargininemia	18,300	1	1:18300
Congenital Hypothyroidism (CH)	10,300	6	1:1700
Congenital Adrenal Hyperplasia (CAH)	10,300	4	1:2600
Galactosemia (GALT)	10,300	1	1:10300
G6PD	8,800	4	1:2200
Overall Incidence Rate			1:540

Incidence Rates - INDIA

Inborn Errors of Metabolism (IEM) – An Indian Perspective, N. B. Kumta, IJP, Volume 72, April 2005. This paper presents the results of a study done at KEM Hospital in Mumbai from 1978 to 2004. The following data is estimated incidence for 24 Million babies born in India.

Metabolic Disorder	Incidence
Congenital Hypothyroidism (CH)	1:2600
G6PD	1:70
Other IEMs	1:1300
Sickle Cell	1:5100

Incidence Rates – US General Population

Virginia Dept of Health – Health Practitioner Manual, March 2006

Metabolic Disorder	Incidence
3-Methylcrotonyl-CoA Carboxylase Deficiency (3MMC or 3MCC)	1:75000
Argininosuccinic Aciduria (ASA)	1:70000
Citrullinemia (CIT)* Citrullinemia Type I	1:57000
Homocystinuria (HCU)	1:150000
Maple Syrup Urine Disease (MSUD)	1:100000
Phenylketonuria (PKU)	1:25000
Carnitine Uptake Defect (CUD) Carnitine Transport Defect (CTD)	1:100000
Long-Chain Hydroxy Acyl-CoA Dehydrogenase Deficiency (LCHADD)	1:75000
Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)	1:25000
Trifunctional Protein Deficiency (TFPD)	1:100000
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCADD)	1:75000
Cobalamin A (cbIA), Cobalamin B (cbIB)	1:100000
Methylmalonic Acidemia (MMA)	1:50000
Methylmalonic CoA Mutase Deficiency (MUT- and MUT°)	1:75000
Beta-Ketothiolase Deficiency (also referred to as Mitochondrial Acetoacetyl CoA Thiolase Deficiency) (BKT)	1:100000
Glutaric Acidemia Type I (GA I)	1:75000
3-Hydroxy-3Methylglutaryl-CoA Lyase Deficiency (HMG)	1:100000
Isovaleric Acidemia/Isovaleric Aciduria (IVA)	1:100000
Multiple CoA Carboxylase Deficiency (MCD)	1:100000
Propionic Acidemia (PA)	1:75000
Biotinidase Deficiency	1:75000
Galactosemia (GALT)	1:50000
Congenital Adrenal Hyperplasia (CAH)	1:25000
Congenital Hypothyroidism (CH)	1:5000
Cystic Fibrosis (CF) Asian	1:31000
Incidence rates for these disorders	1:1350

*Worldwide figures