

**Table 10: Prevalence as of December 2024**

Disorder	Sub group	Screened	Confirmed		Prevalence
<b>Endocrine</b>			<b>8933</b>		
	CH	20,385,390	7963	1:	2,560
	CAH	20,385,390	970	1:	21,016
<b>Amino Acid Disorders</b>		<b>8,446,335</b>	<b>25</b>	<b>1:</b>	<b>337,853</b>
	HCY	8,446,335	2	1:	4,223,168
	MAT	8,446,335	1	1:	8,446,335
	TYR 1	8,446,335	21	1:	402,206
	TYR 2	8,446,335	1	1:	8,446,335
<b>PKU</b>		<b>20,385,390</b>	<b>167</b>	<b>1:</b>	<b>122,068</b>
	PKU - Classical	20,385,390	28	1:	728,050
	PKU - Mild	20,385,390	41	1:	497,205
	PKU - 6PTPS	20,385,390	12	1:	1,698,783
	PKU - Hyperphe	20,385,390	86	1:	237,039
<b>MSUD</b>	MSUD	18,702,314	260	1:	71,932
<b>Organic Acid Disorders</b>		<b>8,446,335</b>	<b>111</b>	<b>1:</b>	<b>76,093</b>
	3MCC	8,446,335	39	1:	216,573
	GA1	8,446,335	50	1:	168,927
	IVA	8,446,335	5	1:	1,689,267
	MMA	8,446,335	8	1:	1,055,792
	MCD	8,446,335	2	1:	4,223,168
	PA	8,446,335	7	1:	1,206,619
	BKD	8,446,335	0	1:	#DIV/0!
<b>OTHERS</b>			<b>330246</b>		
	BTND	8,446,335	15	1:	563,089
	CF	8,446,335	4	1:	2,111,584
	G6PD Def	19,975,189	330227	1:	60
<b>GAL</b>		<b>20,385,390</b>	<b>202</b>	<b>1:</b>	<b>100,918</b>
	GAL - Classical	20,385,390	50	1:	407,708
	GAL - Non Classical	20,385,390	152	1:	134,114

	GAL - Variant	20,385,390	97	1:	210,159
<b>Urea Cycle Disorders</b>		8,446,335	13	1:	649,718
	CIT 1	8,446,335	3	1:	2,815,445
	ASA	7,780,757	10	1:	778,076
<b>Hemoglobinopathies*</b>		7,117,133	2981	1:	2,387
	HbH Disease	7,117,133	2932	1:	2,427
	HbE Disease	7,117,133	290	1:	24,542
	$\beta$ Thalassemia	7,117,133	49	1:	145,248
	HbE, $\beta$ -Thalassemia	7,117,133	27	1:	263,598
	HbE, $\alpha$ -Thalassemia	7,117,133	16	1:	444,821
	HbS, S-Disease	7,117,133	4	1:	1,779,283
	HbS, E-Disease	7,117,133	4	1:	1,779,283
	$\alpha$ Thal Major	7,117,133	3	1:	2,372,378

**Total Confirmed Cases**

**342938**

**PREVALENCE REPORT as of December 2024\***

**1996 - 2024\*\***

Disorders	Screened	Confirmed	Prevalence
<b>Endocrine Disorders</b>			
CH	20,385,390	7,963	1: 2,560
CAH	20,385,390	970	1: 21,016
<b>Amino Acid Disorders</b>			
PKU (28 Classic, 41 Mild, 12 PTPS Def, 86 Hyperphe)	20,385,390	167	1: 122,068
MSUD	18,702,314	260	1: 71,932
OTHER AMINO ACIDS (1 MAT, 21 TYR1, 1 TYR2, 2HCY)	8,446,335	25	1: 337,853
<b>Organic Acid Disorders</b>			
(39 3MCC, 50 GA1, 5 IVA, 8 MMA, 7 PA, 2 MCD)	8,446,335	111	1: 76,093
<b>Urea Cycle Defect</b>			
(1 CIT, 9 ASA)	8,446,335	13	1: 649,718
<b>Galactosemia</b>			
(50 Classical, 152 Non-Classical)	20,385,390	204	1: 100,918
<b>G6PD Def</b>			
	19,975,189	330,227	1: 60
<b>Biotinidase Deficiency</b>			
	8,446,335	15	1: 563,089
<b>Cystic Fibrosis</b>			
	8,446,335	4	1: 2,111,584
<b>Hemoglobinopathies*</b>			
HbH Disease	7,117,133	2,932	1: 2,427
HbE Disease	7,117,133	290	1: 24,542
β Thalassemia	7,117,133	49	1: 145,248
HbE, β-Thalassemia	7,117,133	27	1: 263,598
HbE, α-Thalassemia	7,117,133	16	1: 444,821
HbS, S-Disease	7,117,133	4	1: 1,779,283
HbS, E-Disease	7,117,133	4	1: 1,779,283
α Thal Major	7,117,133	3	1: 2,372,378

\*Hemoglobinopathies is based on newborn screened from 2014 to end of 2023); Fatty Acids Oxidation Disorders are still being reviewed.

\*\*G6PD Def since 2000; MSUD since 2012; AA (other amino acids), UCD, OAD, HGB since 2014; ASA since 2019