Gescreente Neugeborene in Österreich

2017	2018	2019	2020	2021	2022
87.945	85.956	86.952	84.150	86.715	83.189

Entdeckte Fälle

	2017	2018	2019	2020	2021	2022
Phenylketonurie	8	1	7	8	10	6
Hyperphenylalaninämie	5	3	8	5	15	7
Argininämie	-	0	0	1	0	0
Leuzinose	0	0	0	1	0	0
Tyrosinämie Typ I/II	0	0	1	0	1	0
klassische Homocystinurie	0	0	0	0	2	0
Citrullinämie	2	5	0	5	7	1
Argininosuccinat-Lyase-Mangel	0	0	0	0	0	1
Methylmalonazidurie	0	1	0	1	0	0
Propionazidurie	0	2	0	0	1	0
Isovalerianazidurie	0	1	1	0	0	1
Glutarazidurie Typ I	2	2	1	0	0	1
Glutarazidurie TypII, multiple acyl-CoA-Dehydrogenase-Mangel	1	0	0	0	0	0
Medium-Chain-Acyl-CoA-Dehydrogenase-Mangel	5	8	5	8	8	6
Very-Long-Chain-Acyl-CoA-Dehydrogenase-Mangel	1	3	2	3	2	3
Long-Chain 3-OH-Acyl-CoA-Dehydrogenase0Mangel	0	1	0	1	2	0
Carnitintransporterdefekt	1	2	0	0	0	0
Carnitin-Palmitoyl-Transferase-I-Mangel	0	0	0	0	0	0
Carnitin-Palmitoyl-Transferase-II-Mangel	0	0	0	0	0	0
Carnitin-Acylcarnitine-Translokase-Mangel	0	0	0	0	0	0
Cobalamindefekte	0	2	0	0	1	2
Vitamin B12-Mangel	7	56	56	29	44	42
Hypothyreose	35	24	31	24	28	29
Adrenogenitales Syndrom	6	7	5	5	8	11
Biotinidase-Mangel	7	4	6	4	4	5
Galaktose-Stoffwechseldefekte	10	4	4	5	9	6
Zystische Fibrose	21	22	16	17	17	13
Spinale Muskelatrophie					9	10
Angeborene schwere Immundefekte					6	4
Gesamt	111	148	143	117	174	148

Stand Sept. 2023