DEPARTMENT OF PAEDIATRICS
AND ADOLESCENT MEDICINE
MEDICAL UNIVERSITY OF VIENNA

Number of screened newborns in Austria

2014	2015	2016	2017	2018
82.348	84.261	88.060	87.945	85.956

Detected cases

	2014	2015	2016	2017	2018
Phenylketonuria	9	7	4	8	1
Hyperphenylalaninemia	5	8	4	5	3
Maple syrup urine disease	0	0	0	0	0
Tyrosinemia Type I/II	1	0	0	0	0
Hypermethioninemia, Homocystinuria	0	0	1	0	0
Citrullinemia	3	1	2	2	4
Argininosuccinatelyase deficiency	1	1	0	0	0
Methylmalonic aciduria	1	1	1	0	1
Propionic aciduria	1	1	0	0	2
Isovaleric aciduria	0	1	0	0	1
Glutaric acidemia type 1	0	0	4	2	2
Glutaric acidemia type 2, multiple acyl-CoA-dehydrogenase-deficiency	0	0	0	1	0
Medium chain acyl-CoA dehydrogenase deficiency	8	7	6	5	8
Very long chain acyl-CoA dehydrogenase deficiency	2	1	1	1	3
Long chain 3-OH acyl-CoA dehydrogenase deficiency	1	3	2	0	1
Carnitine transporter defect	0	1	0	1	2
Carnitine palmitoyl transferase 1 deficiency	0	1	1	0	0
Carnitine palmitoyl transferase 2 deficiency	0	0	0	0	0
Carnitine-acylcarnitine translocase deficiency	0	0	0	0	0
Cobalamin defects	1	1	0	0	2
Vitamin B12 deficiency	3	3	8	7	56
Hypothyroidism	29	25	32	35	24
Congenital adrenal hyperplasia	4	9	7	6	7
Biotinidase deficiency	13	9	11	7	4
Galactose metabolism defects	6	7	5	10	4
Cystic fibrosis	23	20	18	21	22
Total	111	107	107	111	147

As of June 1, 2019. Diagnoses are available with delay, therefore some numbers may be incomplete yet.