UNIVERSITÄTSKLINIK FÜR KINDER- UND JUGENDHEILKUNDE
MEDIZINISCHE UNIVERSITÄT WIEN

**Gescreente Neugeborene in Österreich**

| 2018 | 2019 | 2020 | 2021 | 2022 | 2023 | 2024 |
|---|---|---|---|---|---|---|
| 85.956 | 86.952 | 84.150 | 86.715 | 83.189 | 77.871 | 77.820 |

**Entdeckte Fälle**

| | 2018 | 2019 | 2020 | 2021 | 2022 | 2023 | 2024 |
|---|---|---|---|---|---|---|---|
| Phenylketonurie | 1 | 7 | 8 | 10 | 6 | 7 | 9 |
| Hyperphenylalaninämie | 3 | 8 | 5 | 15 | 7 | 6 | 12 |
| Argininämie | 0 | 0 | 1 | 0 | 1 | 1 | 1 |
| Leuzinose | 0 | 0 | 1 | 0 | 0 | 0 | 0 |
| Tyrosinämie Typ I/II | 0 | 1 | 0 | 1 | 0 | 0 | 0 |
| klassische Homocystinurie | 0 | 0 | 0 | 2 | 0 | 0 | 0 |
| Citrullinämie | 5 | 0 | 5 | 7 | 1 | 1 | 1 |
| Argininosuccinat-Lyase-Mangel | 0 | 0 | 0 | 0 | 1 | 0 | 1 |
| Methylmalonazidurie | 1 | 0 | 1 | 0 | 0 | 1 | 1 |
| Propionazidurie | 2 | 0 | 0 | 1 | 0 | 0 | 1 |
| Isovalerianazidurie | 1 | 1 | 0 | 0 | 1 | 3 | 1 |
| Glutarazidurie Typ I | 2 | 1 | 0 | 0 | 1 | 0 | 2 |
| Glutarazidurie TypII, multiple acyl-CoA-Dehydrogenase-Mangel | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| Medium-Chain-Acyl-CoA-Dehydrogenase-Mangel | 8 | 5 | 8 | 8 | 6 | 6 | 9 |
| Very-Long-Chain-Acyl-CoA-Dehydrogenase-Mangel | 3 | 2 | 3 | 2 | 3 | 0 | 0 |
| Long-Chain 3-OH-Acyl-CoA-Dehydrogenase-Mangel | 1 | 0 | 1 | 2 | 0 | 3 | 0 |
| Carnitintransporterdefekt | 2 | 0 | 0 | 0 | 0 | 0 | 0 |
| Carnitin-Palmitoyl-Transferase-I-Mangel | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| Carnitin-Palmitoyl-Transferase-II-Mangel | 0 | 0 | 0 | 0 | 0 | 0 | 1 |
| Carnitin-Acylcarnitine-Translokase-Mangel | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| Cobalaminstoffwechseldefekte | 2 | 0 | 0 | 1 | 2 | 2 | 1 |
| Vitamin B12-Mangel | 56 | 56 | 29 | 44 | 42 | 56 | 40 |
| | | | | | | | |
| Hypothyreose | 24 | 31 | 24 | 28 | 29 | 37 | 26 |
| Adrenogenitales Syndrom | 7 | 5 | 5 | 8 | 11 | 8 | 7 |
| Biotinidase-Mangel | 4 | 6 | 4 | 4 | 5 | 10 | 9 |
| Galaktose-Stoffwechseldefekte | 4 | 4 | 5 | 9 | 6 | 3 | 1 |
| Zystische Fibrose | 22 | 16 | 17 | 17 | 13 | 20 | 9 |
| | | | | | | | |
| Spinale Muskelatrophie | | | | 9 | 10 | 14 | 8 |
| Angeborene schwere Immundefekte | | | | 6 | 4 | 3 | 8 |
| | | | | | | | |
| Gesamt | 148 | 143 | 117 | 174 | 149 | 181 | 148 |

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