# Az ORIGIN hordozóság szűrés által vizsgált örökíthető betegségek listája

ORIGIN
HORDOZÓSÁG SZŰRÉS

| | | |
|---|---|---|
| 1. | ACADM | Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency |
| 2. | AGXT | Primary hyperoxaluria type 1 |
| 3. | ARSA | Metachromatic leukodystrophy |
| 4. | ATP7B | Wilson disease |
| 5. | BTD | Biotinidase deficiency |
| 6. | CBS | Homocystinuria due to cystathionine beta-synthase deficiency |
| 7. | CFTR | Cystic fibrosis |
| 8. | DHCR7 | Smith-Lemli-Opitz syndrome |
| 9. | EMD | Emery-Dreifuss muscular dystrophy |
| 10. | FMR1 | Fragile X syndrome |
| 11. | GAA | Pompe disease |
| 12. | GALC | Krabbe disease |
| 13. | GALT | Galactosemia |
| 14. | GBA | Gaucher disease |
| 15. | GJB1 | Charcot-Marie-Tooth disease, X-linked type 1 |
| 16. | GJB2 | Nonsyndromic hearing loss, GJB2-related |
| 17. | GJB6 | Nonsyndromic hearing loss, GJB6-related |
| 18. | GLA | Fabry disease |
| 19. | HADHA | Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency |
| 20. | HBA1 | Alpha thalassemia |
| 21. | HBA2 | Alpha thalassemia |
| 22. | HBB | Sickle cell disease; Beta thalassemia |
| 23. | HEXA | Tay-Sachs disease |
| 24. | MEFV | Familial Mediterranean fever |
| 25. | MMACHC | Methylmalonic aciduria and homocystinuria, cblC type |
| 26. | PAH | Phenylalanine hydroxylase deficiency (Phenylketonuria) |
| 27. | PMM2 | Congenital disorder of glycosylation type 1a |
| 28. | SERPINA | Alpha-1 antitrypsin deficiency |
| 29. | SLC26A2 | Achondrogenesis, type IB; Atelosteogenesis II; Diastrophic dysplasia |
| 30. | SMN1 | Spinal muscular atrophy |

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