Newborn Screening: 1 heel prick test has the potential to diagnose 50 diseases

SIGNIFICANT VARIATIONS EXIST ACROSS COUNTRIES

1 test — **More than 50 diseases**

Disease Abbreviations (used overleaf...)

ALD Adrenoleukodystrophy
ASA Argininosuccinic aciduria
ARG Arginase deficiency
A-T Alpha Thalassemia
BIOPT (BS) Biopterin cofactor biosynthesis deficiency
BIOPT (REG) Biopterin cofactor regeneration deficiency
BKT Deficit of Beta-ketothiolase
B-T Beta thalassemia
BTD Defect of biotinidase
CACT Carnitine / acyl-carnitine translocase deficiency
CAH Congenital Adrenal Hyperplasia
Cbl A Methylmalonic acidemia (CblA)
Cbl B Methylmalonic acidemia (CblB)
Cbl C Methylmalonic Acidemia with Homocystinuria (CblC)
Cbl D Methylmalonic Acidemia with Homocystinuria (CblD)
CF Cystic Fibrosis
CHT Congenital Hypothyroidism
CIT Citrullinemia type I
CIT II Citrullinemia type II (Citrine deficiency)
CPT I Carnitine palmitoyl-transferase (L) deficiency
CPT II Carnitine palmitoyl-trans-ferase II deficiency
CUD Lack of carnitine transport
EXP Short-chain acyl CoA dehydrogenase deficiency
FABRY Fabry Disease
GA I Glutaric acidemia type I
GA2 Glutaric acidemia type II
GAL Galactosemia
GALK Galactokinase deficiency
GNMT Glycine N-methyltransferase deficiency
G6PD Glucose-6-phosphate dehydrogenase
HCU Homocystinuria (CBS deficiency)
HMG 3-Hydroxy 3-methyl glutaric aciduria
H-PHE Benign hyperphenylalaninemia
IBG Isobutyrril-CoA dehydrogenase deficiency
IVA Isovaleric acidemia
LCHAD Long-chain hydroxyacyl CoA dehydrogenase deficiency
MADD Multiplex acyl-CoA dehydrogenase deficiency
MAL Malonic aciduria
MAT Methionine adenosyltransferase deficiency
MCAD Medium-chain acyl CoA dehydrogenase deficiency
MCD Multiple carboxylase deficiency
MLD Metachromatic Leukodystrophy
MMA Vitamin B12 deficiency
MPS I Type I mucopolysaccharidosis
M / SCHAD Short / medium chain 3-OH acyl-CoA dehydrogenase deficiency
MSUD Maple syrup urine disease
MTHFR Homocystinuria due to MTHFR deficiency
MUT Methylmalonic acidemia (Mut)
ORN Hyperornithinemia with Gyrate Atrophy of Choroid and Retina
PA Propionic Acidemia
PKU Phenylketonuria
POMPE Pompe Disease
SAHH Deficit of S-adenosylhomo-cysteine hydrolase
SCD Sickle cell disease
SCID Severe combined immunodeficiency
SMA Spinal Muscular Atrophy
TFP Deficit of the trifunctional protein
TYR I Type I tyrosinemia
TYR II Tyrosinemia type II
TYR III Tyrosinemia type III
VLCAD Very long chain acyl CoA dehydrogenase deficiency
2MBG 2-Methyl butyryl-CoA dehydrogenase deficiency
2M3HBA 2-Methyl 3-hydroxy butyric aciduria
3MGCA 3-methyl glutaconic acids
3MCC Deficit of 3-Methyl crotonyl-CoA carboxyalsi

Country Rankings

	Country	Screening for...
1	Italy	48 Diseases
2	Austria	29 Diseases
3	Portugal	29 Diseases
4	Poland	28 Diseases
5	Hungary	26 Diseases
6	Sweden	26 Diseases
7	Norway	25 Diseases
8	Netherlands	24 Diseases
9	Finland	23 Diseases
10	Germany	20 Diseases
11	Estonia	20 Diseases
12	Czech Republic	19 Diseases
13	Slovenia	19 Diseases
14	Denmark	18 Diseases
15	Slovakia	13 Diseases
16	Switzerland	10 Diseases
17	Belgium	9 Diseases
18	UK	9 Diseases
19	Croatia	9 Diseases
20	Ireland	8 Diseases
21	Spain	7 Diseases
22	France	6 Diseases
23	Latvia	6 Diseases
24	Greece	4 Diseases
25	Lithuania	4 Diseases
26	Bulgaria	3 Diseases
27	Cyprus	2 Diseases
28	Romania	2 Diseases

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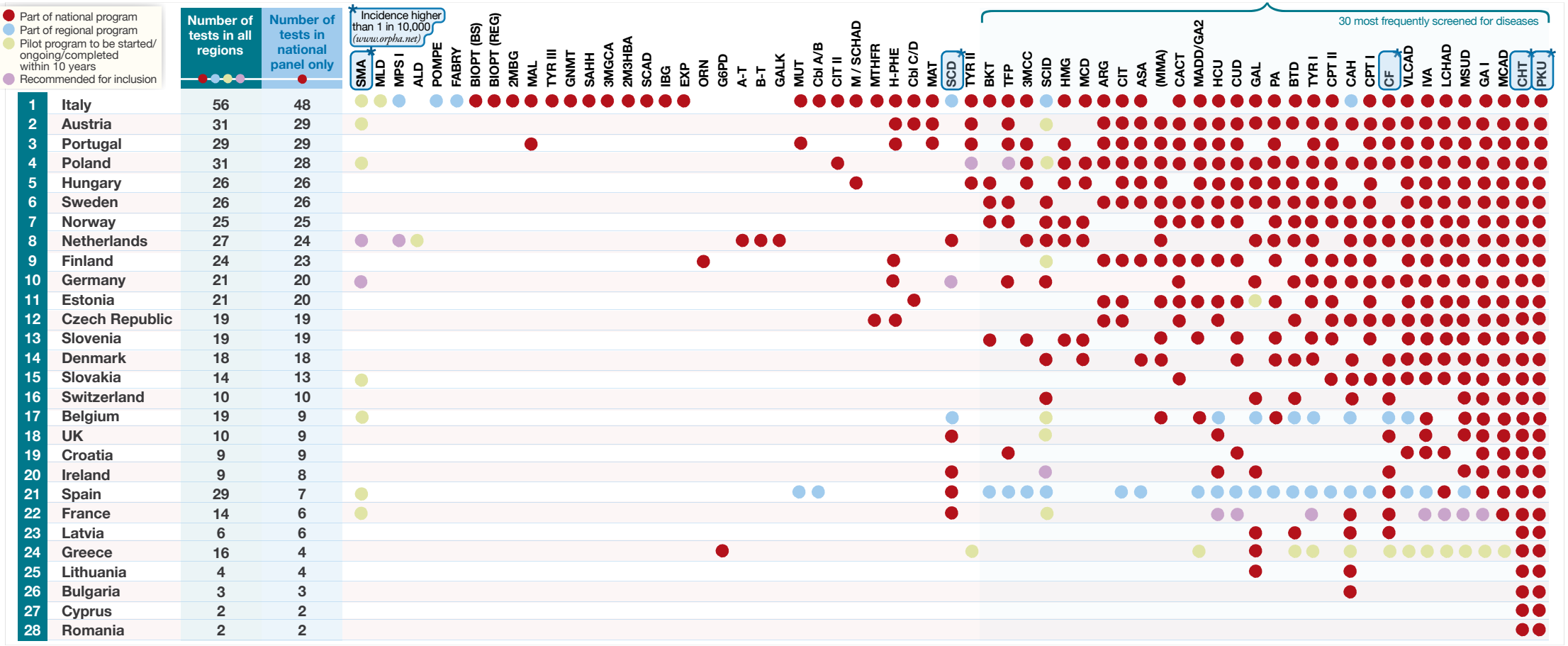

1) Italy • Italian Government - Gazzetta Ufficiale Della Repubblica Italiana – accessible at *https://www.trovanorme.salute.gov.it/norme/renderPdf;spring?s-eriegu=SG&datagu=15/11/2016&redaz=16A08059&artp=1&art=1&subart=1&subart1=10&vers=1&prog=001 (accessed 15/10/2020)*
• Regione Toscana – accessible at: *https://www.regione.toscana.it/-/screening-neonatale (accessed 15/10/2020)*
2) Portugal • Serviço Nacional de Saude (Portugal) - Programa Nacional de Rastreio Neonatal; accessible at: *http://www.insa.min-saude.pt/category/areas-de-atuacao/genetica-humana/programa-nacional-de-diagnostico-precoce/ (accessed 08/10/2020)*
3) Austria • Universitätsklinik für Kinder- und Jugendheilkunde; accessible at: *https://www.meduniwien.ac.at/hp/fileadmin/neugeborenenscreening/pdf/Ansichtsversion_Folder_Neugeborenen_Screening_Finalversion_11.2019.pdf (accessed 08/10/2020)*
4) Poland • Ministry of Health, Poland – accessible at *https://www.gov.pl/web/zdrowie/program-badan-przesiewowych-noworodkow-w-polsce-na-lata-2019-2022 (accessed 05/01/2021)*
5) Hungary • EURODIS, Hungary National Plan & Strategies for Rare Diseases – accessible at *http://download2.eurordis.org/rdpolicy/National%20Plans/Hungary/1.Hungary_RD%20National%20Plan_2013-2020_Hungarian.pdf (accessed 14/10/2020)* • College of Clinical Genetics – accessible at *http://tfsz.pte.hu/docs/protokollok/KL.GENVeleszuletett_anyagcserezavar_I.pdf (accessed 14/10/2020)*
6) Sweden • National Board of Health and Welfare (Socialstyrelsen) – accessible at *https://www.socialstyrelsen.se/globalassets/sharepoint-dokument/artikelkatalog/nationella-screeningprogram/2019-7-6223.pdf (accessed 09/10/2020)* • Karolinksa Institute – accessible at *https://www.karolinska.se/for-vardgivare/karolinska-universitetslaboratoriet/nyheter-kul/2019/08/startdatum-for-screening-for-scid-med-pku-prov/ (accessed 09/10/2020)*
7) Norway • Oslo University Hospital - accessible at *https://oslo-universitetssykehus.no/avdelinger/barne-og-ungdomsklinikken/nyfodtscreeningen/nyfodtscreening (accessed 08/10/2020)*
8) Finland • Sosiaali- ja terveysministeriön - Vastasyntyneiden aineenvaihduntasairauksien seulonnan nykytilanteen selvitys; accessible at *https://julkaisut.valtioneuvosto.fi/bitstream/handle/10024/162289/STM_2020_18_rap.pdf;sequence=1&isAllowed=y (accessed 10/10/2020)*
9) Netherlands • National Institute for Health and Environment Ministry of Health, Welfare and Sport Netherlands; accessible at: *https://www.pns.nl/bielprik/ziekten-die-bielprik-opspoort (accessed 13/10/2020)*
10) Estonia • Tartu University Hospital for Clinical Genetics – accessible at *https://www.kliinikum.ee/geneetikakeskus/vastsuendinute-skriining (accessed 13/10/2020)*
11) Czech Republic • Ministry of Heath, Czech Republic - accessible at *https://www.novorozenecke screening.cz/co-je-novorozenecky-screening (accessed 14/10/2020)*
12) Slovenia • Repič Lampret, B., Remec, Ž.I., Drole Torkar, A., Žerjav Tanšek, M., Šmon, A., Koračin, V., Čuk, V., Perko, D., Ulaga, B., Jelovšek, A.M. and Debeljak, M., Expanded Newborn Screening Program in Slovenia using Tandem Mass Spectrometry and Confirmatory Next Generation Sequencing Genetic Testing- accessible at *https://www.researchgate.net/publication/344553251_Expanded_newborn_screening_program_in_Slovenia_using_tandem_mass_spectrometry_and_confirmatory_next_generation_sequencing_genetic_testing (accessed 08/10/2020)*
13) Germany • The Federal Joint Committee (G-BA) – accessible at *https://www.g-ba.de/downloads/83-691-572/2018_11_22_G-BA_Elterninformation_Erweitertes_Neugeborenen-Screening_bf.pdf (accessed 09/10/2020)* • Daniel, Y., Elion, J., Allaf, B., Badens, C., Bouva, M. J., Brincat, I., ... & Henthorn, J. (2019). Newborn screening for sickle cell disease in Europe. International Journal of Neonatal Screening, 5(1), 15. – accessible at *https://www.mdpi.com/2409-515X/5/1/15/pdf (Sickle cell pilot) (accessed 09/10/2020)*
• DEUTSCHES ÄRZTEBLATT *https://www.aerzteblatt.de/int/archive/article/193172 (Cystic Fibrosis) (accessed 09/10/2020)* • Vill, K., et al (2019). One year of newborn screening for SMA–Results of a German pilot project. Journal of neu-romuscular diseases, 6(4), 503-515. – accessible at *https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6918901/ (SMA) (accessed 09/10/2020)*
14) Denmark • Statens Serum Institut; Sygdomme der indgår i screeningen; accessible at *https://nyfoedte.ssi.dk/medfoedte-sygdomme/sygdomme-der-screenes-for (accessed 08/10/2020)*
15) Slovakia • National Newborn Screening Center of Slovakia – accessible at *http://www.detskanemocnica.sk/rozsireny-novorodenecky-skrining (accessed 13/10/2020)*
16) Switzerland • Kinderspital Zürich –Eleonorenstiftung; Newborn Screening; accessible at: *https://www.neoscreening.ch/en/diseases/ (accessed 13/10/2020)*
17) United Kingdom • Public Health England – Guidance on Blood Spot; Updated 16 August 2019 – accessible at *https://www.gov.uk/government/publications/screening-tests-for-you-and-your-baby/blood-spot (accessed 12/10/2020)* • Public Health England – Newborn blood spot evaluation update — screening for SCID, – accessible at *https://phescreening.blog.gov.uk/2019/10/29/scid-update-bloodspot-screening/ (accessed 12/10/2020)*
• UK National Screening Committee (UK NSC) The UK NSC recommendation on Spinal Muscular Atrophy – accessible at *https://legacyscreening.phe.org.uk/sma (accessed 12/10/2020)*
18) Belgium • Cliniques Universitaire St Luc – Depistage Neonatale, accessible at: *https://www.saintluc.be/laboratoires/activites/biologie-clinique/biochimie/depistage-neonatal.php#:~:text=Pour%20les%20d%C3%A9pister%2C%20des%20tests,buvard%2C%20ou%20carte%20de%20Guthrie (accessed 11/10/2020)* • KCE - Federaal Kenniscentrum - Centre fédéral d'expertise; accessible at: *https://kce.fgov.be/sites/default/files/atoms/files/KCE_267_Bs_D%C3%A9pistage_N%C3%A9onatal_Sanguin.pdf (accessed 11/10/2020)*
19) Croatia • KCB Zagreb - accessible at *https://www.kbc-zagreb.hr/pacijenti-i-posjetitelji/informacije-javnosti-o-novorodenackom-probiru/ (accessed 13/10/2020)*
20) Ireland • Health Service Executive (Ireland), Screening and vaccinations - Heel prick screening; accessible at: *https://www2.hse.ie/screening-and-vaccinations/heel-prick-screening/conditions-the-heel-prick-screens-for/cystic-fibrosis-heel-prick-screening.html (accessed 13/10/2020)*
21) Spain • Castiñeras, D. E., Couce, M. L., Marín, J. L., González-Lamuno, D., & Rocha, H. (2019). Newborn screening for metabolic disorders in Spain and worldwide. Anales de Pediatría (English Edition), 91(2), 128e-1. – accessible at *https://www.sciencedirect.com/science/article/pii/S2341287919301164#bib0150 (accessed 05/10/2020)* • Gobierno de Cantabria – accessible at *https://www.cantabria.es/web/comunicados/detalle/-/journal_-content/56_INSTANCE_DETALLE/16413/1973365 (accessed 05/10/2020)*
• Kotb, M. A., Mansour, L., & Shamma, R. A. (2019). Screening for galactosemia: is there a place for it?. International Journal of General Medicine, 12, 193. – accessible at *https://europepmc.org/article/pmc/6537461 (accessed 05/10/2020)* • Hospital Sant Joan de Deu Barcelona – accessible at *https://metabolicas.sjdhospitalbarcelona.org/noticia/-cataluna-amplia-programa-deteccion-precoz-neonatal-pdpn-22-enfermedades (accessed 05/10/2020)* • Junta de Castilla Y Leon – Consejeria de Sanidad – accessible at *https://www.saludcastillayleon.es/profesionales/en/programas-guias-clinicas/programas-salud/programa-deteccion-precoz-enfermedades-congenitas (accessed 05/10/2020)*
• *https://www.20minutos.es/noticia/4153981/0/la-fe-participa-en-un-estudio-pionero-para-detectar-la-atrofia-muscular-espinal-a-traves-del-cribado-neonatal/ (SMA) (accessed 05/10/2020)* • Centros de Andalucía y Cataluña – accessible at *https://aquas.gencat.cat/web/.content/minisite/aquas/publicacions/2020/avaluacio_cribratge_neonatal_atrofia_espinal_aquas2020.pdf (SMA) (accessed 05/10/2020)*
22) Latvia • Children's Clinical University Hospital Riga – accessible at *https://www.bkus.lv/lv/content/jaundzimusajiem-veiks-skriningu-sesam-slimibam (accessed 10/10/2020)*
23) France • Haute Autorité de santé (France) – accessible at *https://www.has-sante.fr/jcms/p_3149627/fr/depistage-neonatal-quelles-maladies-depister (accessed 13/10/2020)*
24) Greece • Institoúto Ygeías tou Paidioú; The Neonatal Preventive Screening Program in Greece at: *http://ich.gr/en (accessed 08/10/2020)*
25) Lithuania • Ministry of Health, Lithuania – accessible at *https://e-seimas.lrs.lt/portal/legalAct/lt/TAD/6721859095ef11e4b92e9028929aad91 (accessed 13/10/2020)*
26) Bulgaria • Castiñeras, D. E., Couce, M. L., Marín, J. L., González-Lamuno, D., & Rocha, H. (2019). Newborn screening for metabolic disorders in Spain and worldwide. Anales de Pediatría (English Edition), 91(2), 128e-1. – accessible at *https://www.sciencedirect.com/science/article/pii/S2341287919301164#bib0150 (accessed 05/10/2020)*
27) Cyprus • Ministry of Health, Cyprus - accessible at *https://www.moh.gov.cy/Moh/MOH.nsf/All/CD61A07312284C0A422579DC0023AF8A/$file/Strategic%20Plan%20Rare%20Diseases.pdf (accessed 11/10/2020)*
28) Romania • National Institute for Maternal and Child Health, Romania – accessible at *https://insmc.ro/uploads/ftp/pdf/3.prevenireafenilcetonuriei.pdf (accessed 08/10/2020)*

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