

## Bijlage 10: Bronnendocument geboorteprevalentie

CACT: Cornel (2015)/Orphanet  
CPTI: Cornel (2015)/Orphanet  
CPTII: Cornel (2015)/Orphanet  
GALK: Reich (2002)  
GAMT: Pasquali (2016)  
BKT: Cornel (2015)/Orphanet + Gezondheidsraad (2015), Abdelkreem (2016)  
MPS I: UK NSC (2016)  
MMA: Cornel (2015)/Orphanet  
OCTN2: Pilot Denemarken (2016)  
PA: Cornel (2015)/Orphanet  
SCID: Kwan (2014)  
X-ALD: Cornel (2015)/Orphanet

### Referentielijst

- Cornel (2015): Cornel MC. Nieuwe aanbevelingen voor neonatale screening, een rapport van de Gezondheidsraad. Ned Tijdschr Geneesk. 2015;159
- Gezondheidsraad. Neonatale screening: nieuwe aanbevelingen. Den Haag: Gezondheidsraad, 2015; publicatienr. 2015/08.
- [www.orpha.net](http://www.orpha.net)
- Reich (2002): Reich, S., Hennermann, J., Vetter, B., Neumann, L. M., Shin, Y. S., Söling, A., ... & Kulozik, A. E. (2002). An unexpectedly high frequency of hypergalactosemia in an immigrant Bosnian population revealed by newborn screening. *Pediatric research*, 51(5), 598-601.
- Pasquali (2016): Newborn screening for GAMT Deficiency – The Utah Experience. Presentatie beschikbaar:  
[http://southeastgenetics.org/presentation.php/88/Newborn\\_Screening\\_for\\_GAMT\\_Deficiency\\_The\\_Utah\\_Experience](http://southeastgenetics.org/presentation.php/88/Newborn_Screening_for_GAMT_Deficiency_The_Utah_Experience)
- UK NSC (2016). The UK NSC recommendation on mucopolysaccharidosis type I. Beschikbaar via: <https://legacyscreening.phe.org.uk/mps1>
- Pilot Denemarken (2012). Lund, A. M., Hougaard, D. M., Simonsen, H., Andresen, B. S., Christensen, M., Dunø, M., ... & Larsen, N. (2012). Biochemical screening of 504,049 newborns in Denmark, the Faroe Islands and Greenland—Experience and development of a routine program for expanded newborn screening. *Molecular genetics and metabolism*, 107(3), 281-293.
- Kwan (2014(. Kwan, A., et al (2014). Newborn screening for severe combined immunodeficiency in 11 screening programs in the United States. *JAMA*, 312(7), 729-738.
- Abdelkreem (2016). Abdelkreem E, Otsuka H, Sasai H, Aooyama Y et al. Beta-Ketothiolase Deficiency: Resolving Challenges in DiagnosisJournal of Inborn Errors of Metabolism & Screening 2016, Volume 4: 1–9.

