

## Newborn screening for galactosemia

### Summary

**Objectives:** To evaluate the effectiveness and cost-effectiveness of newborn screening for galactosemia in order to improve the nationwide newborn screening programme in Estonia.

**Methodology:** To describe the newborn screening programmes elsewhere in the world and gather evidence on their effectiveness and cost-effectiveness a literature review was conducted. The current situation in Estonia was described using unpublished data from the laboratory that organises existing newborn metabolic screening programme. Budget impact analysis was carried out to determine the effect of adding galactosemia screening to the present nationwide screening programme on healthcare payers' budget.

**Results:** There is an existing nationwide newborn metabolic screening programme in Estonia. Newborns are already screened for 19 inborn metabolic diseases and congenital hypothyroidism. Since 2015 there has also been a pilot screening programme for galactosemia. For all of the screened diseases, including galactosemia, the samples are gathered with dried bloodspots and analysed from the same testcard. The screening laboratory has already made arrangements in its job management to improve 2<sup>nd</sup> tier testing and rearrange the testing procedure so that galactosemia tests could be analysed first. In current analysis, the additional cost of screening an annual birth cohort of 14,000 newborns was estimated at €50,000 – 60,000 per year.

**Conclusions:** Since galactosemia is a rare disease, there are only a few publications on the effectiveness and cost-effectiveness of the disease. Based on the information gathered and analysed, newborn screening would be the best possible approach to early detection of galactosemia. There is an existing nationwide newborn metabolic screening programme in Estonia and adding the galactosemia screening to that programme should be considered.

**Citation:** Kiisk E, Joost K, Kiivet R-A. *Galaktoseemia sõeluuring Eestis*. Tartu: Tartu Ülikooli peremeditsiini ja rahvatervishoiu instituut; 2020.