

Summary

Preimplantation genetic testing for monogenic abnormalities

Objective: To evaluate the cost-effectiveness and budget impact of preimplantation genetic testing for monogenic abnormalities (PGT-M) in Estonia compared with natural conception, to inform decisions on routine public funding.

Methods: Systematic literature searches were conducted in PubMed for clinical guidelines, evidence on effectiveness and safety, and economic evaluations of PGT-M. In addition, international reimbursement practices were reviewed. A meta-analysis of single-arm studies was performed to estimate clinical pregnancy and live birth rates following *in vitro* fertilisation (IVF) with PGT-M. Safety evidence from meta-analyses comparing preimplantation genetic testing plus IVF versus IVF alone was summarized. A de novo cost-effectiveness model and a five-year budget impact analysis were developed from the Estonian Health Insurance Fund perspective, assuming up to three PGT-M cycles per couple.

Results: Eleven guidelines from seven organizations recommend considering PGT-M for serious monogenic diseases with a clear genetic cause, substantial impact on life expectancy or quality of life, and limited treatment options. Public funding for PGT-M is available in several countries, typically with restrictions on indications and number of cycles.

No comparative studies directly assessed the effectiveness of PGT-M plus IVF versus natural conception. In 19 single-arm studies, pooled clinical pregnancy and live birth rates per IVF cycle with PGT-M were 51% (95% CI 45–58%) and 44% (95% CI 38–50%), respectively. Safety outcomes were largely similar between preimplantation genetic testing plus IVF and IVF alone, although preterm birth was slightly more frequent with the former (OR 1.12, 95% CI 1.03–1.21). No evidence compared the combined safety of PGT-M and IVF with natural conception.

Seven cost-effectiveness studies showed generally favourable results for PGT-M, though many had methodological limitations. In the Estonian model, PGT-M with IVF was cost-saving over a 10-year horizon, yielding an average gain of 0.1 quality-adjusted life years (QALY) per woman and reducing costs, primarily by preventing births of children with severe monogenic diseases and reducing pregnancy terminations. Results were robust in most sensitivity analyses at a €20,000/QALY threshold.

The budget impact analysis, assuming 10 eligible couples annually, estimated an additional cost of €77,900 in year one with PGT-M, decreasing to €27,300 by year five, with net savings emerging from year seven onward due to avoided treatment costs. Compared with current partial cross-border funding, routine funding would add only modest biopsy- and transportation related costs.

Conclusions: Publicly funded PGT-M could be cost-saving and have a limited short-term budget impact in Estonia when restricted to clearly defined indications and a limited number of cycles.

Citation: Alloja J, Pallasma E, Muru K, Ehrenberg A, Juus E, Jürisson M. Embrüote siirdamiseelne testimine monogeense haiguse suhtes, tervisetehnoloogiate hindamise raport TTH79. Tartu: Tartu Ülikooli peremeditsiini ja rahvatervishoiu instituudi tervisetehnoloogiate hindamise keskus; 2025.