



IQWiG Reports – Commission No. S15-02

Newborn screening for severe combined immunodeficiency (SCID screening)¹

Extract

¹ Translation of the key statement of the final report S15-02 *Screening auf schwere kombinierte Immundefekte (SCID-Screening) bei Neugeborenen* (Version 1.0; Status: 16 November 2016). Please note: This document is provided as a service by IQWiG to English-language readers. However, solely the German original text is absolutely authoritative and legally binding.

Publishing details

Publisher:

Institute for Quality and Efficiency in Health Care

Topic:

Newborn screening for severe combined immunodeficiency (SCID screening)

Commissioning agency:

Federal Joint Committee

Commission awarded on:

30 July 2015

Internal Commission No.:

S15-02

Address of publisher:

Institut für Qualität und Wirtschaftlichkeit im Gesundheitswesen
Im Mediapark 8
50670 Köln
Germany

Phone: +49 221 35685-0

Fax: +49 221 35685-1

E-mail: berichte@iqwig.de

Internet: www.iqwig.de

This report was prepared in collaboration with external experts.

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External experts

- Michael Borte, Clinic for Paediatric and Adolescent Medicine, St. Georg Hospital gGmbH, Leipzig, Germany
- Tim Mathes, Monika Becker, Dawid Pieper, Institute for Research in Operative Medicine, University Witten/Herdecke, Cologne, Germany

IQWiG thanks the external experts for their collaboration in the project.

IQWiG employees

- Anne Rummer
- Elke Hausner
- Lina Rodenhäuser
- Stefan Sauerland
- Wiebke Sieben

Keywords: Neonatal Screening, Transplantation, Severe Combined Immunodeficiency, Benefit Assessment, Systematic Review

Key statement

Research question

The aim of this investigation is to assess the benefit of newborn screening for severe combined immunodeficiency (SCID screening) in combination with curative treatment (allogeneic bone marrow or stem cell transplantation) compared with no SCID screening. The focus of the investigation was on patient-relevant outcomes.

Conclusion

There is a hint of a benefit for SCID screening in combination with treatment to prevent infections, which is followed by curative treatment.

A dramatic effect in favour of an earlier versus a later start of treatment to prevent infection is described for the outcome of mortality. The data from the studies on diagnostic quality were not sufficient to calculate sensitivity and specificity. The positive predictive value (PPV) for SCID screening by the determination of T-cell receptor excision circles (TREC) from filter card blood shows that the test for SCID is basically suitable for identifying children with SCID. It is unclear how many children are not detected.

These results are based on 3 non-randomized comparative intervention studies at the start of treatment as well as 5 studies on diagnostic quality, which were combined using a linked evidence approach. All studies show a high risk of bias.

An ongoing French comparative intervention study of the screening chain, in which data on sensitivity and specificity are also to be collected, suggests that results will be obtained to answer the question of the benefit of SCID screening; data will be collected up to June 2018.

The full report (German version) is published under

<https://www.iqwig.de/de/projekte-ergebnisse/projekte/nichtmedikamentoesse-verfahren/s-projekte/s15-02-screening-auf-schwere-kombinierte-immundefekte-scid-screening-bei-neugeborenen.6805.html>