Newborn screening outcomes of mixed blessing

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In this part of the project we use two 'categories' of test outcomes: false positives and positive test results for a metabolic disease that was not tested. Objectives of the study are to evaluate these categories of test outcomes by…

Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Metabolic and nutritional disorders congenital
Study type	Observational non invasive

Summary

ID

NL-OMON33650

Source
ToetsingOnline

Brief title
Newborn screening unexpected outcomes

Condition

- Metabolic and nutritional disorders congenital
- Inborn errors of metabolism

Synonym
false positive newborn screening results of screened inborn errors of metabolism in The Netherlands 2007-2009

Research involving
Human

Sponsors and support

Primary sponsor: TNO Leiden Center for Child Health and Pediatrics
Source(s) of monetary or material Support: Centre for Society and Genomics (CSG) via Centre for Medical Systems Biology (CMSB)

Intervention

Keyword: False Positive Result, Neonatal Screening, Quality of Life

Outcome measures

Primary outcome

1. Change in anxiety level and health perception of their child in parents with

false positive outcomes.

2. Views and experience of parents confronted with another disease than

screened for.

Secondary outcome

1. Balancing benefits of the outcomes of mixed blessing against uncertainties

and worries in terms of welfare, autonomy

and knowledge.

2. Implications for screening criteria and informed consent and follow-up

procedures.

Study description

Background summary

Newborn screening is conducted on almost all newborns in The netherlands during the first week of life. Until recently, blood was tested for three severe congenital disorders which can be treated before clinical symptoms arise: phenylketonuria (PKU), congenital hypothyroidism (CHT) and adrenogenital syndrome (AGS). However, new technologies such as tandem mass spectroscopy (MS/MS) and DNA micro-array have been developed to cover a wide range of diseases that can be traced using the heel prick screening. Based on the advisory report published by the Health Counsil the newborn screening has been expanded with 14 disorders since the first of January 2007, leading to an estimated increase of 80-90 detection per year, primarely caused by positive test results for sickle-cell disease. The other disorders involve smaller numbers of detected cases. The expanded programme has been set up according to

the recommendations and the screening criteria proposed by the Health Counsil in the past. Important criteria are: that a good test method is available, screening and treatment are available for every patient and the screening should be in the interest of the child. In light of these criteria, the expansion leads to a number of unexpected or unwanted test outcomes. Although unexpected or unwanted the results may still be valued positively as far as these lead to more certainty and knowledge of the medical condition of the child. This may play a role in unexpacted or unwanted outcomes of newborn screening and can lead to implications for newborn screening criteria, informed consent criteria, and follow-up procedures.

Study objective

In this part of the project we use two 'categories' of test outcomes: false positives and positive test results for a metabolic disease that was not tested. Objectives of the study are to evaluate these categories of test outcomes by empirical research. The results of the empirical study will be used for recommandations for screening.

Study design

This empirical and retrospective study will be carried out by means of a questionnaire and semi-structured interview, evaluating the views and experiences of parents with non-intended screening outcomes of mixed blessing. The results of this study will be used for ethical analysis and evaluation: implications for newborn screening criteria, informed consent procedures, and follow-up procedures. Parents are asked to participate by their pediatrician. Controls are selected by means of date of birth (RIVM).

Study burden and risks

Burden: questionnaires: 30 minutes; interviews: 2 hours.
Risks: none.
Benefit: express and share experiences, influance on informed consent and long term follow-up procedures.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adults (18-64 years)
Elderly (65 years and older)

Inclusion criteria

False positive newborn screening result 2007-2009
Normal result of the newborn screening 2007-2009

Exclusion criteria

Abnormal result newborn screening otherwise (i.e. iatrogenic, prematurity)

Study design

Design

Study type:	Observational non invasive
Intervention model:	Other
Allocation:	Non-randomized controlled trial
Masking:	Open (masking not used)

Control:	Active
Primary purpose:	Health services research

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	01-01-2010
Enrollment:	1750
Type:	Actual

Ethics review

Approved WMO

Date:	24-09-2009
Application type:	First submission
Review commission:	METC Leids Universitair Medisch Centrum (Leiden)

Study registrations

Followed up by the following (possibly more current) registration

No registrations found.

Other (possibly less up-to-date) registrations in this register

No registrations found.

In other registers

Register	ID
CCMO	NL25677.058.09