*Cochrane Database of Systematic Reviews - - Cochrane Review*

# Preconception risk assessment for thalassaemia, sickle cell disease, cystic fibrosis and Tay-Sachs disease

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## Study design (if review, criteria of inclusion for studies)

randomised controlled trials (RCTs) and quasi-randomised controlled trials (quasi-RCTs).

## List of included studies (0)

No eligible trials were identified.

## Participants

Women of reproductive age (aged 16 to 50 years old) who are carriers for thalassaemia, sickle cell disease, cystic fibrosis or Tay-Sachs disease, with or without partners, accessing any healthcare services which includes hospitals and community-based healthcare settings. Community-based healthcare settings include family or general practices, community health centres, community health services, community or outpatient clinics and ambulatory care services. If studies contain both eligible and ineligible participants, they will be included if data on eligible participants can be extracted.

## Interventions

systematic preconception genetic risk assessment for thalassaemia, sickle cell disease, cystic fibrosis or Tay-Sachs disease, in any healthcare setting. We define systematic preconception genetic risk assessment as a package of risk assessment including one or more of these components: â€¢ family history assessment; â€¢ assessment of ethnicity background; â€¢ genetic carrier testing; â€¢ genetic carrier screening.

## Outcome measures

Primary outcomes 1. Reproductive outcomes in women who are carriers of thalassaemia, sickle cell disease, cystic fibrosis or Tay-Sachs disease identified during or after pregnancy i) number of infants born with genetic conditions ii) number of infants born with congenital anomalies iii) number of infants born with low birth weight iv) number of infants born prematurely 2. Decisions about future conception and pregnancy in women who are carriers for thalassaemia, sickle cell disease, cystic fibrosis or Tay-Sachs disease i) number of women or couples who would make use of prenatal diagnosis ii) number of women or couples who would make use of prenatal diagnosis and consider termination of pregnancy if the child is affected iii) number of women or couples who would consider pre-implantation genetic diagnosis and in vitro fertilization iv) number of women or couples who would conceive using donated gametes v) number of women or couples who would consider adoption vi) number of women or couples who would refrain from having any children

## Main results

No RCTs of preconception genetic risk assessment for thalassaemia, sickle cell disease, cystic fibrosis and Tayâ€•Sachs disease are included. A trial identified earlier has published its results and has subsequently been listed as excluded in this review.

## Authors' conclusions

As there are no RCTs of preconception genetic risk assessment for thalassaemia, sickle cell disease, cystic fibrosis, or Tayâ€•Sachs disease included in either the earlier or current versions of this review, we recommend considering potential nonâ€•RCTs studies (for example prospective cohorts or beforeâ€•andâ€•after studies) for future reviews. While RCTs are desirable to inform evidenceâ€•based practice and robust recommendations, the ethical, legal and social implications associated with using this trial design to evaluate the implementation of preconception genetic risk assessment involving carrier testing and reproductive autonomy must also be considered. In addition, rather than focusing on single geneâ€•byâ€•gene carrier testing for specific autosomalâ€•recessive conditions as the intervention being evaluated, preconception expanded genetic screening should also be included in future searches as this has received much attention in recent years as a more pragmatic strategy. The research evidence for current international policy recommendations is limited to nonâ€•randomised studies.

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**See also**

Hussein N, Henneman L, Kai J, Qureshi N. Preconception risk assessment for thalassaemia, sickle cell disease, cystic fibrosis and Tayâ€•Sachs disease. Cochrane Database of Systematic Reviews 2021, Issue 10. Art. No.: CD010849. DOI:10.1002/14651858.CD010849.pub4. Accessed 25 October 202110.1002/14651858.CD010849.pub3

**Keywords**

Genetic Testing; Heterozygote Detection; diagnostic procedures; non pharmacological intervention - diagn; Truth Disclosure;