Other Reviews - - Other Review

Effectiveness of preconceptional and prenatal cystic fibrosis carrier screening: a systematic review.

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

systematic review. Primary and secondary studies on adults (16 years and older), with no clinical indication or genetic risk, eligible for genetic testing for CF carrier status.

Participants

General population. Adults of reproductive age

Interventions

Genetic testing for cystic fibrosis for CF carrier screening

Outcome measures

Attitude to screening, uptake of screening offered, informed reproductive choices.

Main results

A total of 3,326 records were screened and 292 potentially eligible full-text publications assessed. The review included 71 publications, corresponding to 3 reviews, 40 cohort studies (11 comparative, 29 single-arm), and 6 model studies, published between 1992 and 2021 (median 1998). Only one study compared screening or no screening. This study suggested an association between carrier screening and a lower incidence of CF. Comparative studies examined different approaches for invitation and testing, i.e., settings, target population (individuals/couples, prenatal/preconceptional), how invitations are organized (primary care/maternal hospitals), and format and content of the pre-test information. However, no firm conclusions can be drawn on the impact of these features on informed reproductive choices, uptake, and attitude, because of the limitations of the evidence collected.

Authors' conclusions

The broad heterogeneity of the studies, methodological weaknesses, and the limited transferability of the results mean there is still uncertainty about the effectiveness of preconceptional and prenatal CF carrier screening in the general population.

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See also

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Keywords

Adult; carrier status; computer programs; Counseling; Family Therapy; Genetic Predisposition to Disease; Genetic Testing; Heterozygote Detection; non pharmacological intervention - diagn; non pharmacological intervention - psyco-soc-edu-org; screening; training; Psychoeducation; Systemic interventions; diagnostic procedures; non pharmacological intervention - genetic& reprod;