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Utility of preconception carrier testing to prevent autosomal recessive diseases

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Objective

To assess the rate of autosomal recessive and X-linked disorders that can be prevented by using preconception carrier testing.

Methods

Carrier testing enables to determine the reproductive risk associated with specific autosomal recessive (AR) and X-Linked (XL) conditions in the general population. Recent evidence shows that offering carrier screening to all couples, regardless of their ethnicity, effectively and equitably identifies those at-risk, although there is no agreement on the number of diseases to be screened. In this study, we assessed the rate of diseases that could have been prevented by disclosing the carrier status of monogenic diseases with a carrier frequency higher than 1/200.

Results

Among 274 fetuses studied in our center, 88 (32%) causative diagnoses were revealed by next generation sequencing, including 34 monogenic disorders with AR inheritance and 3 with XL inheritance. Seven (19%) of those had a carrier frequency >1/200 in the general population: Smith-Lemli-Opitz syndromes (n=2), Niemann-Pick disease, polycystic kidney disease, short-rib thoracic dysplasia, Ellis-van Creveld syndrome and X-linked syndromic intellectual developmental disorder.

Conclusion

Carrier testing could predict seven of the diagnoses with AR or XL monogenic inheritance, accounting for 8 % of all diagnoses. Our results demonstrate a limited clinical utility of reproductive carrier screening in order to prevent the appearance of autosomal recessive and X-linked disorders.