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REVIEW

Carrier screening for Beta-thalassaemia: a review of international practice

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β-thalassaemia is one of the most common single-gene inherited conditions in the world, and thalassaemia carrier screening is the most widely performed genetic screening test, occurring in many different countries. β-thalassaemia carrier screening programmes provide a unique opportunity to compare the delivery of carrier screening programmes carried out in different cultural, religious and social contexts. This review compares the key characteristics of β-thalassaemia carrier screening programmes implemented in countries across the world so that the differences and similarities between the programmes can be assessed. The manner in which thalassaemia carrier screening programmes are structured among different populations varies greatly in several aspects, including whether the programmes are mandatory or voluntary, the education and counselling provided and whether screening is offered pre-pregnancy or antenatally. National and international guidelines make recommendations on the most appropriate ways in which genetic carrier screening programmes should be conducted; however, these recommendations are not followed in many programmes. We discuss the implications for the ethical and acceptable implementation of population carrier screening and identify a paucity of research into the outcomes of thalassaemia screening programmes, despite the fact that thalassaemia screening is so commonly conducted.
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INTRODUCTION

Screening for genetic diseases aims to reduce the burden of these disorders on individuals by identifying those at increased risk, thereby enabling individuals to receive information about their personal health, future health and/or potential health of their offspring.[1] Population carrier screening programmes aim to identify asymptomatic carriers of recessive conditions, so that they are informed and understand their reproductive risks and options.[2–4] If both members of a couple are found to be carriers of an autosomal recessive condition, they have a one in four chance of having an affected child. Carrier screening programmes are conducted throughout the world. National and international guidelines recommend that genetic screening should not be compulsory for any individual or population; they also recommend that appropriate information be provided to individuals before testing to enable informed decision making about genetic screening, and that genetic screening be accompanied by counselling.[2–7] An ethical concern arises when people are uninformed about the consequences of medical tests and interventions undertaken.[7] However, at present, there is no consensus on the most suitable way by which to deliver genetic screening programmes such that they meet these recommendations, and it is likely that the provision of these is affected by cultural, social and religious beliefs. A genetic screening programme that provides the opportunity to compare different approaches taken to screen populations around the world is β-thalassaemia carrier screening, as this is one of the most common single-gene inherited conditions in the world.[8–10]

Thalassaemias are haemoglobinopathies that are characterized by a decrease in the production of globin chains, which results in microcytic anaemia.[1–5] The two major types of thalassaemia are α-thalassaemia and β-thalassaemia. α- and β-thalassaemia occur when there is a reduction in α-globin chains and β-globin chains, respectively.[11,12]

Programmes offering screening for β-thalassaemia heterozygotes (carrier screening) have been available for many years.[4,13] The majority of β-thalassaemia carriers have reduced mean corpuscular volume and mean corpuscular haemoglobin levels in the standard full blood examination (FBE).[8] A person's carrier status can be confirmed by haemoglobin electrophoresis[8] and/or high-performance liquid chromatography.[14]

Almost 70 000 infants are born with β-thalassaemia worldwide each year and 270 million people are carriers of haemoglobinopathies.[15,16] β-thalassaemia is most commonly present among populations in all Mediterranean countries, as well as in Southeast Asia, India, Africa, Central America and the Middle East.[14,15,17,13] However, because of migration, the carrier rate of β-thalassaemia is increasing in countries that previously had low prevalences. β-thalassaemia causes severe, blood-transfusion-dependent anaemia in people who are homozygous or compound heterozygous for mutations in the β-globin gene (β-thalassaemia major).[17] The main complication of this therapy is that frequent transfusions lead to iron overload, as humans have no mechanism to excrete excess iron.[17,18] Iron overload can cause liver cirrhosis and cardiomyopathy. Iron chelation therapy is used to increase iron excretion, which prolongs the life of individuals with thalassaemia.[19]

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Table 1 Thalassaemia screening programs conducted around the world (ordered from time of screening program implementation)

Author	*District /country*	*Delivery, time period*	*Antenatal/ premarital*	*Mandatory/voluntary*	*Informed about screening/ consent given*	*Education*	*Counselling*
Theodoridou[65]	Greece (whole country)	Universal, since 1973	Premarital and antenatal	Voluntary	Informed consent	Educational programs at school, in the armed forces, maternity clinics and through the mass media. Carriers informed and given a leaflet.	Carrier couples offered counselling
Angastiniotis and Hadjiminas[25], Cowan[26]	Cyprus (whole country)	Universal, since 1973	Preconceptional (planning a wedding or pregnancy) or antenatal	Quasi-mandatory by church among Greek Cypriots; Mandatory by Government among Turkish Cypriots	Informed	Public education through mass media, talks in schools, clubs, communities, printed information	Counselling offered to couples or individuals concerned
Cao[66,67]	Sardinia, Italy	Outreach community and hospital testing, since 1975	Premarital or antenatal (couples/adults of child-bearing age)	Voluntary	Informed consent	Posters, information booklets, group discussions, presentations, through mass media	Counselling provided to carriers and carrier couples
Lena-Russo *et al*[55]	Marseille region, France	High schools, 1978–1985	Premarital	Voluntary	Informed consent	Educational session	Counselling provided for carriers and their families
Scriver *et al*[46], Mitchell *et al*[27]	Montreal, Canada	Neighbourhood health centres and high schools, 1979–1992	Premarital (high school and community centres)	Voluntary	Informed consent	Pre-screening instruction sessions and letters, pamphlets and posters distributed	Carriers and carrier couples offered counselling
Zlotogora *et al*[28]	Israel (whole country)	Offered to Arab and Druze populations, as well as Jews originating from Iran, Iraq, Syria, Kurdistan, Mediterranean countries and Asiatic countries of the former USSR, since 1980	Premarital or antenatal	Voluntary	Informed consent	Explanatory movie and pamphlet	Genetic counselling offered in local clinics before and after screening to carriers and carrier couples
Greengross *et al*[52]	London, England	Central Middlesex Hospital antenatal clinic, since 1985	Antenatal	Voluntary	No	No	Counselling for carriers and their partners and for carrier couples
Firdous[43]	The Maldives (whole country)	Universal, since 1992	Premarital (people aged 12–35)	Voluntary	Informed consent	Awareness program (print materials and school education)	Counselling provided to everyone when receiving test results.
Chern[68]	Taiwan (whole country)	Universal, since 1993	Antenatal	Voluntary	No	No	No
Liao *et al*[54]	Guangdong Province, China	Guangzhou Maternal and Neonatal Hospital, Jan 1993- Dec 2003	Antenatal	Voluntary	No	Education programs for carrier couples	Counselling for husbands of carriers, and carrier couples
Samavat and Modell[24], Abolghasemi *et al*[14], Karimi *et al*[22], Khorasani *et al*[23]	Iran (whole country)	Universal since 1997, since 1991 in Sari, since 1995 in Southern Iran	Premarital (couples that want to register for marriage)	Mandatory	Informed	Mass media, information booklets, educational programs, classes in high school and for young men in the military. Carrier couples receive information at counselling to make informed decisions	Counselling for carrier couples. Pregnancy termination and prenatal diagnosis became legal in 1998. Prenatal diagnosis and abortion are now discussed during counselling. Couples can attend as many sessions as they want until they can make an informed decision about what to do
Colah *et al*[30]	Mumbai city, India	Wadia Maternity Hospital, 1997–2003	Antenatal	Voluntary	Informed consent	Posters displayed and leaflets given to the women	Carriers were counselled with their husbands, and carrier couples were offered prenatal diagnosis

Table 1 (Continued)

Author	*District /country*	*Delivery, time period*	*Antenatal/ premarital*	*Mandatory/voluntary*	*Informed about screening/ consent given*	*Education*	*Counselling*
Sirdah[69]	Palestinian District, Gaza Strip	High schools, 1998	Premarital	Voluntary	Informed consent	60 minute lecture given a day before testing	No
Tarazi *et al*[9]	Palestinian District, Gaza Strip	Universal, since 2000	Premarital (for all engaged couples)	Mandatory	Informed	Awareness campaign	Carrier couples receive counselling. Advised to cancel marriage. If couple wants to continue with marriage, they sign a declaration to say that they are aware that they are both carriers
Acemoglu[70], Canatan[71]	Turkey (33 provinces)	Universal, since 2002	Premarital (couples wishing to get married)	Mandatory since 2005	Informed	Public education	Counselling provided for carrier couples
AlHamdan *et al*[20], Al Sulaiman[72]	Saudi Arabia (whole country)	Universal, since 2003	Premarital (for all couples applying for a marriage license)	Mandatory	Informed	No	Counselling for carrier couples. Carrier couples advised to separate as prenatal diagnosis and pregnancy termination not widely practised
NHS Sickle Cell and Thalassaemia Screening Programme[33], Old[34], Locock and Kai[32]	England (whole country)	Universal, since 2004	Antenatal	Voluntary	Informed consent (should occur)	Information provided for carriers. Information should be provided to women before testing so that an informed decision can be made	Counselling can be arranged for carriers to make an informed decision about prenatal diagnosis

Thalassaemia carrier screening is arguably the mostly widely performed carrier screening test. In this study, we review the different β-thalassaemia carrier screening programmes conducted throughout the world and compare key characteristics of the delivery of these programmes. A literature search was conducted using the following databases: Medline, Pubmed, Psychinfo and Cinahl. Peer-reviewed articles that describe β-thalassaemia screening programmes conducted throughout the world were included in this review. It is likely that there are many other thalassaemia screening programmes conducted in other countries not mentioned; however, they have not yet been described in a peer-reviewed journal.

Thalassaemia carrier screening programmes

Thalassaemia carrier screening programmes identified in the published literature are outlined in Table 1. They can be divided into programmes that are largely mandatory and those that are voluntary. Programmes can also be divided by the timing of testing in relation to pregnancy, being either pre-pregnancy or in the early stages of pregnancy. There are more options available to a couple if screening has occurred before conception. A couple can decide to end their relationship, adopt a child, use donor egg or sperm, conceive through *in-vitro* fertilization using pre-implantation genetic diagnosis or they can choose to go ahead with prenatal diagnosis after conception and elect to terminate the pregnancy if the foetus is affected with β-thalassaemia major.

Mandatory screening

World Health Organization guidelines, published in 1998, stated that no compulsory genetic testing should be carried out.[3] Nevertheless, some countries including Iran, Saudi Arabia, Palestinian Territories and Cyprus have laws in place making premarital screening for haemoglobinopathies mandatory for all couples before they are given approval to get married.[9,20–26] This raises ethical questions, as couples are unable to voluntarily decide for themselves whether they would like to undergo genetic screening. Thalassaemia carrier screening has been included as part of an existing mandatory premarital blood test in Iran since 1991.[22–24] This form of mandatory premarital thalassaemia screening also began in the Gaza Strip in 2000[9] and in Saudi Arabia in 2003.[20] Carrier couples receive advice on the options available to them, one of which is cancellation of marriage, and they can then decide whether to marry each other.[20,22,24] A quasi-mandated premarital β-thalassaemia carrier screening programme began in Cyprus in the early 1980s.[26] Cyprus has one of the highest carrier rates in the world, with an estimated carrier rate of one in seven.[26] Among Greek Cypriots, premarital screening is mandated by the Cypriot Orthodox Church.[26] Couples wanting to get married by the Church are required to be screened and counselled by the Thalassaemia Centre and be issued with a certificate.[26] In all these countries, mandatory screening is practised in a confidential manner, and even though premarital screening is mandatory, carrier couples still have the right to get married if they wish. These countries believe that this method of screening is the most effective to decrease the incidence of thalassaemia.[20]

Voluntary screening

Other haemoglobinopathy screening programmes are offered on a voluntary basis. Voluntary programmes have been conducted in high schools, as well as before pregnancy and antenatally. A β-thalassaemia genetic screening programme was conducted in high schools in Montreal.[27] During education sessions before the screening, information was provided to the high school students, and the students were

given time to provide consent for the testing.[27] In this study, the students become aware of their carrier status generally before they have settled with a partner with whom they plan to have children. Thalassaemia screening is also offered in Israel on a voluntary basis, where people are able to undergo screening after being informed about the test, either before or at the beginning of a pregnancy.[28]

Antenatal screening is another voluntary option conducted in many countries.[29] An antenatal β-thalassaemia carrier screening programme was implemented in India, in which pregnant women provide informed consent at their first antenatal visit before a blood test is conducted.[30] However, in some countries, particularly those with a lower prevalence of thalassaemia, antenatal screening is offered in an *ad hoc* manner.[31,32] It is often the practitioner's decision whether to offer screening. In Australia, there is no consistent screening protocol/policy for practitioners to follow, other than requesting an FBE early in a woman's pregnancy. Previously, the United Kingdom did not have a consistent model for screening and/or counselling;[31] however, a universal antenatal screening programme for haemoglobinopathies has recently been developed, and this programme is offered to all pregnant women.[32–34] All pregnant women undergo an FBE, and in high prevalence areas of the United Kingdom, additional testing[32] through a high-performance liquid chromatography analysis is also performed routinely.[33,34] All women with a low mean corpuscular haemoglobin level will also undergo a high-performance liquid chromatography analysis for diagnosis of β-thalassaemia carrier status.[34]

It is an open question whether participation in this screening process is voluntary. There is a significant lack of public awareness of thalassaemia carrier screening in many countries, including the United Kingdom, United States of America and Australia. In some countries, thalassaemia carrier screening is carried out in the majority of pregnant women, as part of a routine, initial FBE during early pregnancy.[35,36] The FBE is carried out for a number of different reasons, including diagnosing anaemia, as well as thalassaemia carrier screening.[31,35] The test is offered as a matter of course during pregnancy and some women see it as a mandatory, routine blood test.[32,37] Most women are not aware that they are being screened for thalassaemia until they are found to be a carrier,[31,35,36] unless a discussion occurs about what may be revealed from the FBE results.[38] In a number of these thalassaemia screening programmes, women are tested without actually being informed about the test.[36] Even if these screening programmes are not considered mandatory for pregnant women, it is likely that many examined individuals are unaware that it is occurring.

Ahmed and colleagues surveyed British pregnant women who had undergone carrier testing about their awareness of the testing, before implementation of a national antenatal screening programme.[37] It was found that 77.4% of the women had not been informed about thalassaemia carrier screening, and 85.8% of these women wanted to have been informed.[37] Although the new UK-screening programme advises that women should provide informed consent, Locock and Kai found that people in the United Kingdom were still not aware that they were being screened, and women were often shocked when presented with the results.[32] They revealed that many people in the United Kingdom would prefer to be tested for haemoglobinopathy carrier status before conception, or even before choosing a partner,[32] which gives the women many more options regarding conception and the birth of a child with thalassaemia. In the United States of America, pregnant women were also found to have a lack of awareness of genetic testing, and if they had undergone carrier testing, they often could not recall undergoing the test.[39] Ormond and colleagues found that many pregnant women feel overwhelmed about the numerous tests offered to them, and they do not have a clear understanding about what any of the tests are designed to detect.[39] It should be noted that antenatal screening for thalassaemia often occurs in the context of the first antenatal visit, at which time a considerable amount of information about testing and care during pregnancy is provided[40] and recall may be poor if the information is not judged to be of particular relevance.[41]

Informed consent for thalassaemia carrier screening

A number of different guidelines have recommended that informed consent should be a requirement for all genetic screening programmes.[2–4] Population thalassaemia carrier screening is offered after obtaining informed consent in certain countries that have a higher prevalence of thalassaemia, particularly Mediterranean and Asian countries. The communities are educated about thalassaemia before individuals make a decision about whether they will undergo carrier testing. The community education programmes in these countries increase the population's awareness of thalassaemia.[42] In the Maldives, an awareness programme is offered to the community to provide public education about thalassaemia, before pre-conceptional screening is offered and consent is required to undergo testing.[43]

One major barrier to obtaining informed consent for genetic testing is the lack of knowledge about genetic disorders among many health professionals.[44] Current information should be available to all health professionals so that they are capable of informing their patients.[29] Other barriers to gaining informed consent for every genetic test offered include time consumption and cost. Even though genetic screening tests themselves may be inexpensive, the human resources required to provide education and counselling for every genetic test can be very costly.[45] As new genetic screening tests become available, the time and money required to offer pre-test information and obtain informed consent must be considered,[29] as even though informed consent is highly recommended, realistically it may not always be practical to obtain informed consent from each individual for every genetic test offered.[35,39,44]

Despite the recent implementation of a universal haemoglobinopathy screening programme in the United Kingdom, thalassaemia screening programmes in countries such as the United Kingdom and Australia often involve indirect genetic testing, as the test is usually presented as a routine FBE in pregnant women and little or no information about the screening test for thalassaemia carrier status is presented to the women.[32] Ahmed and colleagues found that most women who knew that they had been carrier screened for thalassaemia believed that the test was a routine test and they did not have a choice with regard to the test being conducted.[37] This form of carrier screening is carried out in a manner very different from other carrier screening programmes, such as those for cystic fibrosis and Tay Sachs disease, conducted in high schools, which involve obtaining informed consent before testing.[35,38] There are no specific guidelines on how to obtain consent for indirect genetic testing.[36]

Ahmed and colleagues in the United Kingdom showed that women believe that being informed about thalassaemia screening is a completely separate issue from providing consent to being tested.[37] Most women wanted to be informed about the test; however, they did not necessarily feel that they had to give consent to the test.[37] The majority of women in that study showed trust in their health professionals, and hence believe that their midwives and doctors know what is best for them, and are happy to have the screening tests recommended; however, they would prefer to be informed about the tests conducted.[37] Many women did not want to make the decisions about

carrier testing, and preferred their doctors to decide for them.[37] Similar findings were shown in a study conducted by Locock and Kai, in which women indicated that they would have preferred to be informed about the thalassaemia carrier screening; however, consent was not seen as an important issue.[32]

Education about thalassaemia carrier screening

Knowledge and understanding about thalassaemia carrier screening can improve if public education is provided. Some thalassaemia carrier screening programmes educate participants before the screening process; however, the participants are all educated in slightly different ways. Successful community-wide education programmes are conducted in countries with high carrier rates, such as Greece, Italy and Cyprus.[42] Armeli found that 85% of the study participants in Italy had heard of thalassaemia, whereas only 19% of Italian–Americans living in the United States of America had heard of it.[42] Native Italians showed a much higher level of knowledge of thalassaemia compared with Italian–Americans.[42] Education has been provided through mass media, lectures presented to the general public, training of health professionals, as well as posters and pamphlets in marriage registry offices and medical clinics.[42]

Education is offered in countries in which premarital thalassaemia carrier screening is mandatory, such as Iran, as even though people are unable to make their own decision about whether to undergo testing, it is believed that they should still be informed about the test that they must undergo. In Iran, classes on thalassaemia are conducted in high schools and are also conducted for young men in the military.[24] Public education has also been carried out in Iran through mass media, annual public education programmes and information booklets written by the Youth Thalassaemia Group.[22]

Attitudes about thalassaemia carrier screening

Overall, there is a positive attitude towards thalassaemia screening in the few studies examining this question. Before a high school β-thalassaemia carrier screening programme began in Quebec, Canada, 88% of the people informed about the programme at community centres and high schools believed that the screening should begin as soon as possible.[46] Once the programme began, 95% of high school students tested (carriers and non-carriers) approved the screening experience.[46] Locock and Kai found that women who had undergone antenatal carrier screening in the United Kingdom were overall glad to be made aware of their carrier status.[32]

A positive attitude has also been shown in two countries in which screening is currently not routinely offered. Gilani and colleagues questioned medical practitioners, lawyers, politicians and parents of children affected by thalassaemia in Pakistan, to determine people's attitudes about thalassaemia carrier screening.[47] Over 95% of the parents and 90% of the doctors supported genetic screening; however, only one-third of politicians were in favour of screening.[47] Thalassaemia screening has not yet been introduced in Sri Lanka; however, 96% of people agreed that at least one person in a marriage should undergo thalassaemia screening.[48]

Outcomes of thalassaemia carrier screening

Reduced incidence. A major outcome of thalassaemia carrier screening is a reduction in the incidence of thalassaemia.[25,49–51] The main factors that can lead to a reduction in disease incidence is an increase in the uptake of prenatal diagnosis and use of reproductive technologies to prevent the births of affected children,[52] as well as a decrease in marriages between carriers. In many countries, a large majority of the affected foetuses detected are terminated.[27,30,53–56]

The incidence of β-thalassaemia has decreased significantly after the introduction of screening programmes. The voluntary carrier screening programme, which began in Sardinia, Italy, in 1975 reduced the incidence of β-thalassaemia from 1:250 to 1:4000 in 1995.[50] All of the carrier couples originally identified as carriers in a high school screening programme in Montreal, Canada, chose prenatal diagnosis, and all affected foetuses were terminated.[27] Therefore, because of this carrier screening programme, no one screened has given birth to an affected child, which has caused a 95% decrease in the incidence of β-thalassaemia in that region.[27,57] Similar results were seen in Marseille, France, where 86% of partners of carriers identified many years earlier in a high school screening programme were carrier tested, and all carrier couples requested prenatal diagnosis and all of the affected pregnancies identified were terminated.[55] A premarital screening programme began in Cyprus in 1973 and the number of affected births decreased from 51 in 1974 to 8 in 1979.[25] The incidence continued to decrease after the screening programme further developed into a mandatory screening programme in the early 1980s, with only five affected births occurring between 1991 and 2001 and no affected births occurring between 2002 and 2007.[58] Before carrier screening began in Cyprus, difficulties were experienced in obtaining enough blood supply for the treatment of all affected individuals[59] and this would have become a larger problem if the incidence continued to rise.[60] This decrease in incidence has benefitted those affected with thalassaemia, as the demand for blood has decreased, therefore improving the supply of treatment in Cyprus.

An antenatal thalassaemia screening programme in Taiwan resulted in a significant reduction in the incidence of β-thalassaemia. Before the programme, about 20 children with thalassaemia were born every year.[61] Seven years later, the incidence had decreased to only three to six affected children being born every year.[61] Another antenatal screening programme has been conducted in Guangdong, China, for 11 years.[54] Over 95% of the carrier couples identified underwent prenatal diagnosis and only one child with β-thalassaemia has been born, because of a misdiagnosis during prenatal diagnosis.[54] All of the affected foetuses detected in the screening programme were terminated.[54]

Most potential parents in the United Kingdom will choose to terminate their pregnancy if the foetus is affected by β-thalassaemia.[53] Greengross and colleagues showed that 80% of β-thalassaemia-affected births were prevented by the antenatal screening programme conducted at Central Middlesex Hospital between 1986 and 1995.[52,62] It is expected that the current universal antenatal carrier screening programme in the United Kingdom will further decrease the incidence of β-thalassaemia.

Some countries have not offered prenatal diagnosis to couples, as pregnancy termination is forbidden on religious grounds. Before prenatal diagnosis and pregnancy termination for high-risk couples was legalized in Iran, about half of the carrier couples identified by the premarital screening programme proceeded to marry each other, and the other half separated.[24] The separation of these high-risk couples helped to reduce the incidence of β-thalassaemia.[24] In contrast, the incidence of β-thalassaemia has changed little in Saudi Arabia,[20] even though it is mandatory for couples to participate in the premarital β-thalassaemia carrier screening programme.[20] Almost 90% of the high-risk couples identified within 2 years married each other after being screened,[20] and prenatal diagnosis and pregnancy termination is not widely practised. The incidence of thalassaemia in Saudi Arabia may decrease, however, as pre-implantation genetic diagnosis is becoming a popular alternative.

Anxiety. A negative outcome that can be caused by β-thalassaemia carrier screening is an increase in anxiety levels. Carriers are often initially anxious about their positive test results.[46] As expected, carriers who show the highest anxiety levels are those whose partner is also found to be a carrier, particularly if the woman is pregnant when the carrier status is identified.[35] Anxiety in women who are identified as carriers during antenatal screening has been shown to decrease when they are reassured that their partners are not carriers.[63]

CONCLUSION

Guidelines and recommendations about the conduct of carrier screening programmes, developed by organizations such as the World Health Organization[3,5,6] and Nuffield Council on Bioethics,[2] include recommendations such as a requirement of informed consent before the conduct of a genetic test[2] and that no compulsory genetic testing should occur.[3] In contrast to these recommendations, this review shows that it is not uncommon for thalassaemia screening programmes to be mandatory and/or not notify individuals that they are being tested unless they are found to be carriers. Clearly, screening programmes have been adapted to meet the needs of different communities and seem to reflect acceptable cultural practice.

Although the incidence of β-thalassaemia after screening may be documented as an outcome of screening, the differing cultural and medical contexts of each country and every population mean that there is unlikely to ever be one best strategy to screen a population.[30] However, programmes can be evaluated against the objectives that they set themselves and against the assumptions – or 'program logic'[64] – that underlie decisions about delivery, such as the timing of provision of screening, the extent to which education is offered and whether consent is important. Programme evaluation theory stresses the importance of establishing a programme that is consistent with the documented preferences, beliefs and behaviours of the target community, as well as clearly establishing the objectives at the outset, and then evaluating a programme's process and outcomes on the basis of these.

No one strategy can meet the needs of every country and every population. However, incorporating programme evaluation can determine whether specific needs of each particular country or community are being met and provide valuable data that may inform others who are considering the implementation of carrier screening.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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