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PROGRAM REPORTS

A collaborative genetic carrier screening model for the British Ashkenazi Jewish community

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Abstract

We present a unique model of a British genetic carrier screening programme for individuals with Ashkenazi Jewish ancestry that exemplifies a partnership between a publicly funded healthcare service (the NHS) and a charity, Jnetics. This model provides affordable access to carrier screening for severe autosomal recessive diseases increased in this community. Prior to the development of this programme, the British healthcare system only provided Tay Sachs' screening for this community, leaving them at higher risk of having a child with a serious autosomal recessive disease. The Jnetics screening programme is promoted through community and social media campaigns, involves educational outreach, a pre-test genetic counselling service by a dedicated NHS-based genetic counsellor, saliva-based DNA testing, comprehensive reporting and, where required, post-test genetic counselling. The charity raises funds to subsidise the screening. In 6 years, the model has been successfully implemented in hospital and community settings and in schools and universities, aiming to reach those pre-conception. In response to the COVID-19 pandemic, the programme adapted by offering genetic screening virtually and has subsequently expanded in its outreach. Furthermore, the screening panel is currently being expanded to include other conditions increased in the Ashkenazi and also the Sephardi and Mizrahi Jewish communities. An example of innovation and accessibility, providing free screening to all students and disadvantaged individuals, the programme aims to provide a model that can potentially be adopted by other genetically at-risk communities.

Keywords Carrier · Screening · Ashkenazi · Jewish · Recessive

Ashkenazi Jews comprise an estimated 95% of the 290,000 British Jews and have a higher prevalence of several inherited autosomal recessive conditions compared to the general population—a prime example of the founder effect following reproductive isolation. It is estimated that 1 in 5 Ashkenazi Jews are a carrier for at least one of 11 heritable conditions (Klugman and Gross 2010). Most of these conditions are fatal in early childhood, are associated with significant morbidity and have a large emotional and psychological impact on families (Warsch et al. 2014).

Carrier screening in the USA has been routinely offered to the Ashkenazi Jewish community for over 15 years for a panel of autosomal recessive conditions, following recommendations from the American College of Obstetricians and Gynaecologists and American College of Medical Genetics (Shao et al. 2015). Since 1983, a carrier screening programme, Dor Yeshorim, has been available to the ultra-orthodox Jewish community where individuals cannot access their genetic results but are 'matched' with potential marriage partners based on their carrier results (Dor Yeshorim 2021). This, however, would not be relevant to the majority of British Jews who are not ultra-orthodox.

In 1999, the UK National Screening Committee approved Ashkenazi Jewish carrier screening for Tay Sachs disease under the publicly funded National Health Service (NHS). However, screening for other at-risk genetic conditions was not offered. This resulted in a population of Ashkenazi Jews of reproductive age who remained at risk of having a child with a preventable serious genetic disorder.

We report on the creation and expansion of the first British community-based genetic screening programme, uniquely set up as a partnership between the NHS and the

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UK charity, Jnetics. Established in 2009, Jnetics aims to increase awareness of Jewish genetic disorders, improve access to their support services and facilitate access to professional testing services (Jnetics 2021). In 2014, it launched its community-based screening programme, providing educational outreach and access to screening for 9 autosomal recessive diseases (Table 1). These 9 were selected based on a high diagnostic specificity, affordability and their severity and carrier frequency in the Ashkenazi Jewish population.

Jnetics screening initially took place in community centres and synagogues, later moving into a clinical setting with the establishment of the "Jnetics Clinic" in 2017. In 2017, Jnetics also introduced screening into Jewish secondary schools in the form of the student 'GENEius programme'—offering 'screening days' to students aged 16–18 in 8 schools across London and Manchester. The GENEius programme has been expanded to university students, with screening events taking place at 4 universities to date. The cost to have screening at the 'Jnetics Clinic' is subsidised by the charity at £250; however, the charity covers the cost for all students and individuals who do not have the means.

The Jnetics screening process begins online where participants can access detailed information about the screen and self-refer through an online portal and questionnaire. At the appointment, genetic screening information and individual risk calculations based on family history are discussed, as well as the availability of family planning options for 'carrier couples' (carriers of the same condition). After gaining consent, a saliva sample is collected using a saliva test kit and then sent to the laboratory. Results are released alongside a comprehensive letter within 8–10 weeks and all carriers have the opportunity to speak to a genetic counsellor.

Table 1 Nine Ashkenazi Jewish autosomal recessive genetic diseases screened for by Jnetics with reported carrier frequencies in the Ashkenazi Jewish population (Scott et al. 2010) and in the Jnetics cohort. Genetic testing for the nine conditions was carried out in an NHS laboratory using a panel test for 65 mutations using Amplification Refractory Mutation System-PCR

Genetic condition	Carrier frequencies	
	Ashkenazi Jewish	Jnetics[y]
Cystic fibrosis	1 in 23	1 in 35
Tay-Sachs disease	1 in 27	1 in 37
Familial dysautonomia	1 in 31	1 in 48
Canavan disease	1 in 55	1 in 45
Glycogen storage disease type 1a	1 in 64	1 in 95
Mucolipidosis IV	1 in 89	1 in 95
Fanconi anaemia type C	1 in 100	1 in 95
Niemann-Pick disease type A	1 in 115	1 in 333
Bloom syndrome	1 in 134	1 in 142

[y]Jnetics carrier frequencies include those of full and part Ashkenazi Jewish ancestry

For pregnant participants of the Jnetics Clinic, results are available within 5–6 weeks, and if indicated, the partners of carriers are referred to their local genetic service for further counselling and testing or they can choose to attend the Jnetics Clinic. Virtual screening using postal saliva kits and counselling over the phone was made available in the Jnetics clinic in 2018 to allow greater access to the screening programme.

The school GENEius programme provides extra support through a mandatory educational pre-screening assembly, with the information provided to parents by email. Students consent to have testing but can give permission for results to be shared with parents. The effectiveness of the programme is monitored through anonymised pre and post-education session questionnaires.

The screening process has always relied on the unique collaboration between Jnetics and the NHS. This enables the charity to focus on strategic over-view, organisation and promotion of the screening programme, with the NHS having oversight and control of clinical delivery and governance, including confidential storage of genetic results. Uniquely, the charity pay the NHS for the service of a dedicated genetic counsellor and administrator contracted through a regional NHS genetics service. From 2014 to March 2020, genetic testing took place in an NHS-accredited laboratory on a private basis. Since April 2020, testing has taken place in a private CLIA-certified and CAP-accredited laboratory, which will shortly deliver an expanded genetic screening panel for Jnetics.

The charity promotes its programme through the Jewish press and media, community organisations and synagogue groups. Religious leaders also promote the screening to engaged couples under their auspices. Of interest, a number of participants who had a screening with Dor Yeshorim sought repeat testing through Jnetics in order to access their results.

A large social media campaign was created with the launch of the GENEius programme, having proven to be successful in accessing student populations for Jewish genetic screening (Foxler et al. 2015). More recently, publicity has taken a unique turn with students volunteering to promote the programme themselves on site as 'GENEius Campus Representatives'.

Data from the GENEius programme and the Jnetics clinic from their inception in 2017 until March 2020 shows that 2581 individuals had a screening, of which 80% were screened through the GENEius programme and 20% in the clinic. Fifty-five percent of participants were female and 45% were male. Sixty-two per cent of participants had four Ashkenazi Jewish grandparents and 8% reported a family history of one of the nine conditions. Of the 2581 individuals, 382 (15%) individuals were identified as carriers for at least one of the nine disorders. Of these individuals, 358

were identified as carriers for one disease, 23 were carriers for two diseases and one was a carrier for three diseases. The carrier frequencies are summarised in Table 1. As the programme screened people with both full and part Ashkenazi Jewish ancestry, the carrier rates are less than reported in the literature (Scott et al. 2010). Twenty percent of people attending the Jnetics clinic (103 individuals) were expecting a child at the time of screening. If identified as a 'carrier couple', they were able to access prenatal testing. Four 'carrier couples' were identified pre-conception and they had the additional option of preimplantation genetic diagnosis.

Prior to COVID-19, 17% of all Jnetics clinic participants chose to have a virtual screening session by phone. Jnetics adapted rapidly and dynamically to the COVID-19 pandemic, converting to an exclusive virtual screening programme, consisting of pre-test counselling sessions by phone and home-delivered saliva test kits. Furthermore, the UK's first virtual university genetic screening event in November 2020 saw 250 students participate from 58 colleges and universities, reflecting the increased capacity and accessibility of virtual screening compared to traditional screening. Jnetics has recently expanded its screening panel to include other conditions increased in both the Ashkenazi Jewish community and the Sephardi and Mizrahi Jewish community (of Middle Eastern, Spanish or North African ancestry). Plans are also in place to explore the possibility of community-wide BRCA gene screening.

Jnetics has demonstrated that an effective partnership between a charity-run genetic screening programme and the NHS can be established for a minority community in the UK. Since 2018, Jnetics has been involved with Breaking Down Barriers (funded primarily by the Sylvia Adams' Charitable Trust), a network that supports families from marginalised communities affected by genetic disorders, ensuring they have equal access to healthcare. Sharing Jnetics' unique and accessible screening model aims to highlight a successful example of screening to other at-risk communities in Britain and worldwide.

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Data availability Fully anonymised data was available from the Jnetics charity.

Code availability Not applicable.

Declarations

Ethics approval and consent to participate This article does not contain any studies with human or animal subjects performed by any of the authors.

Conflict of interest Monica Ziff volunteers for the Jnetics charity as a screening advisor. Juliette Harris is a non-paid trustee of the charity, Jnetics.

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