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Journal of Community Genetics

ISSN 1868-310X

J Community Genet
DOI 10.1007/s12687-016-0278-0

Springer

J Community Genet
DOI 10.1007/s12687-016-0278-0

PROGRAM REPORTS

Community engagement and education: addressing the needs of South Asian families with genetic disorders

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Received: 14 April 2016 / Accepted: 12 August 2016

Abstract Consanguineous marriage is common among the South Asian heritage community in the UK. While conferring social and cultural benefits, consanguinity is associated with an increased risk of autosomal recessive disorders and an increase in childhood death and disability. We have previously developed a genetic service to address the needs of this community. We report the extension of this service to include community-based initiatives aimed at promoting understanding of genetic issues related to consanguinity and improving access to genetic services. Our approach was to develop integrated clinical, educational and community engagement initiatives that would be sustainable on a long-term basis. The service provided for South Asian families by a specialist genetic counsellor was extended, and a series of genetics education and awareness sessions were provided for a diverse range of frontline healthcare workers. Two community genetic outreach worker posts were established to facilitate the engagement of the local South Asian population with genetics. The education and awareness sessions helped address the lack of genetic knowledge among primary health care professionals and community workers. Engagement initiatives by the genetic outreach worker raised awareness of genetic issues in the South Asian community and families affected by autosomal recessive disorders. All three elements of the extended service generated positive feedback. A three-stranded approach to addressing the needs of consanguineous families affected by autosomal recessive disorders as recommended by the World Health Organisation is suggested to be an acceptable, effective and sustainable approach to delivery of service in the UK.

Keywords Consanguinity · Autosomal recessive disorders · Genetic counselling · South Asian · Community engagement

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Introduction

Consanguinity, which refers to reproductive couples being close blood relatives, is prevalent in many communities in North Africa, the Middle East and West Asia, where it accounts for between 20 and over 50 % of all marriages (Bittles 1990, 2012; Hamamy 2012). Consanguineous marriages confer social and cultural benefits (Modell 2002), continue to be practised and are common among migrant communities in the UK originating from countries and regions where consanguinity is traditional. The Pakistani heritage community is the largest community in the UK to favour consanguinity (Darr and Modell 1988). The impact of consanguinity on prevalence of congenital abnormalities, autosomal recessive disorders, death and disability in infancy and childhood in this community has been the subject of previous publications (Ahmad 1994; Bundey and Aslam 1993; Sheridan et al. 2013) and is recognised as a significant health burden.

The World Health Organisation (WHO) recommends a family-centred approach, which "recognises that consanguineous marriage is an integral part of cultural and social life in many areas and that attempts to discourage it at the population level are undesirable and inappropriate" (Alwan and Modell 1997). Emerging opinion suggests that community

Published online: 10 September 2016

engagement should be an integral component of any genetic service development (WHO 1985; Salway et al. 2012)

Blackburn with Darwen (BwD) is a co-terminous Unitary Authority and Clinical Commissioning Group (CCG) in North West England. Approximately 30 % of the population is Muslim and of South Asian heritage, split almost equally between Indian and Pakistani (Census 2011). The number of non-Muslim South Asian persons in BwD is small (around 1/40th of the total South Asian population), mainly Buddhist, Hindu and Sikh. The highest incidence of consanguinity is in the Pakistani community within BwD (around 75 %). However, where the Indian population practice consanguinity, this has resulted in similar profiles of autosomal recessive disorders. In South Asian families that are not overtly consanguineous, the practice of intra-biraderi marriage is thought to be prevalent but difficult to measure.

In the period 2002–2010, BwD had the highest mortality rate in the country for children aged 1–17 years (annual average: 27.6/100,000 BwD; 16.5/100,000 UK), and in 2008–2010, the highest infant mortality rate in the country (annual average: 8.0/1000 live births BwD; 4.6/1000 UK) (Local Authority Child Health Profiles. http://www.chimat.org.uk/profiles/static accessed 28 Feb 2016). Analysis of the Child Death Overview Panel (CDOP) data indicated that 41 % of BwD deaths under age 18 years are due to chromosomal, congenital and genetic disorders (unpublished CDOP data).

A clinical database of autosomal recessive disorders in children aged under 16 years developed and maintained in BwD since 2002 indicates a prevalence of 2.6/1000 in White British children, compared with 24.3/1000 in South Asian children (unpublished data). We have identified a broad range of autosomal recessive disorders in BwD, many of which are amenable to diagnosis by mutation analysis, enabling genetic counselling and carrier testing of extended family members as well as prenatal or preimplantation genetic diagnosis. However, we have previously reported that there is little awareness of inherited disorders within the South Asian population in BwD and have identified that lack of knowledge, poor communication, language barriers and stigma are all factors inhibiting access to genetic services (Khan et al. 2010).

A variety of innovative services and community engagement initiatives have been piloted in areas with large South Asian populations, but few have been evaluated or are receiving ongoing funding (Salway, personal communication 2015). The Enhanced Genetic Services Project undertaken in Birmingham involved initiatives in primary care with haemoglobinopathy screening, the identification and review of families with autosomal recessive disorders known to the Clinical Genetics service with the provision on enhanced counselling and molecular testing and the delivery of educational activities in the community and with health care professionals An evaluation report was published in 2014 (Alberg et al. 2014) setting out recommendations for potential service development.

Following a Department of Health funded service development initiated in 2002, we established and evaluated a hospital-based genetic counselling service in BwD for parents and extended relatives in consanguineous families where a child has been diagnosed with an autosomal recessive disorder (Khan et al. 2010). However, we recognised the need to take a broader approach to improve access to genetic services for the South Asian community and complement existing clinical genetic services with community education and engagement. The primary aim of our current initiative was to establish a sustainable model for service provision and community engagement.

Methods

There were three strands to our service development as shown in Table 1

The first strand of our service development was the initiation of a clinical service led by a specialist genetic counsellor, which adopted a family-centred approach, focusing on identifying high risk consanguineous families in which at least one member was already affected by an autosomal recessive disorder. Affected adults and children were offered genetic investigation and counselling, followed by cascade counselling

Table 1 The three strands of our extended service development

Start date	Development	Purpose
2007 onwards	Family-centred genetic counselling service for consanguineous families	Provide genetic services for high risk families: identify affected individuals, trace family members, offer genetic counselling and genetic testing
2012 onwards	Training for frontline professionals and community workers	Address poor understanding of genetics: develop skills and confidence for other health care workers to provide preliminary genetic information and make appropriate referrals to specialist genetic services
2014 onwards	Establish community genetic outreach worker posts	Develop community engagement activities to raise awareness of genetic issues, increase uptake of services and support affected families

and the offer of genetic testing to members of the extended family. This service has been evaluated (Khan et al. 2010) and identified that key factors in the success of this service development were the provision of a locally based genetic counselling service that addressed potential language and cultural barriers to accessibility and acceptability. This service has subsequently been extended to incorporate genetic counselling clinics held in the local antenatal clinic in conjunction with midwives and obstetricians.

Questionnaire feedback was obtained from families who had attended the specialist genetic counsellor clinic. The questionnaires were written in English and were followed up by a telephone call from another member of the service who went through the questions and documented responses during the telephone contact if the individual was willing to do so.

Strand two of the development was to provide genetic educational opportunities for frontline health and social care workers. Half-day training sessions were set up, open to all interested health care community workers, including midwives, health visitors, practice nurses, early years workers and social workers. These were advertised in Children's Centres, by email to all contacts in voluntary and children services and by word of mouth by colleagues. Teaching sessions were also provided for hospital and community paediatricians as part of their existing educational meetings and shorter sessions for GPs at their surgeries. To ensure that other key agencies were aware of this initiative, presentations were also made to the Chair of the Clinical Commissioning Group (CCG), at CCG locality meetings, to Child Death Overview panel members and to Voluntary Community Faith (VCF) organisations.

The objectives of these teaching sessions were to

- Integrate genetic perspectives into established health promotion activities
- Raise awareness and understanding of genetic and cultural issues related to consanguinity
- Empower front line workers in the health and children's services to initiate discussions on genetic issues
- Enable community health workers to identify families at increased genetic risk
- Raise awareness of the availability and scope of local genetic services
- Develop referral pathways for families requiring genetic services
- Define professional roles and responsibilities

After each educational session, participants were given a feedback questionnaire. Free form comments were also invited

The third strand of our initiative aimed to establish effective community engagement. The unique elements of this approach were the employment, training and supervision of two genetic outreach workers in the community (one Urdu speaking and one an experienced screening midwife) and the development of strong working partnerships between the Regional Genetic Service and key agencies including Public Health, Primary Health Care and the VCF sector. The role of the outreach workers was to support a holistic approach to meeting families' genetic, health and social needs. Their remit was to act as a link between the families and the diverse network of medical and support services available and to engage with the extended family members and wider community networks.

Results

Impact on referrals to the dedicated genetic counsellor clinic

Genetic services in Blackburn have been provided by consultant clinical geneticists for many years. Since the establishment of the dedicated genetic counsellor clinics in 2007, approximately an extra 25 families a year have been seen by the genetic counsellor. Following the community education programme, a further 20 additional families (each comprising several relatives who required counselling) were referred to the newly established genetic counsellor clinic held monthly in the local antenatal department in 2015. Of these families, 15 were from midwives, 2 from paediatricians and 3 from the outreach workers. It remains to be seen how many referrals will be received in the future from GPs following the educational sessions.

Questionnaires requesting feedback about their contact with the service were sent to 37 consecutive individuals who had attended the specialist genetic counsellor clinic over a 12-month period (34 South Asian, 1 Polish, 2 white British). Five completed questionnaires were returned by post, and 20 responses were obtained during the subsequent phone call. This was not a formal evaluation, rather an exercise to gain feedback to help monitor service provision and highlight any changes that needed to be made.

The majority of families (19; 76 %) understood why they had been referred to the genetic clinic, but only 10 (40 %) had been aware of genetic services beforehand. Ten (40 %) families did not know what to expect from the clinic appointment, but the remainder expected to be given information and to discuss options for future pregnancies. All 25 respondents said they understood the genetic information given to them and that this was useful. Twenty-three respondents (93 %) found the appointment helpful, and 22 (88 %) felt they would be able to explain to their relatives what the genetic implications were for them. Seventeen respondents felt less worried after the consultation, five more worried and three felt no different. There are limitations of these

results due to the small number of participants and the collection of responses by phone call.

Comments made by respondents included

"Helped me understand why children are ill in my family. It all makes sense now"
"Knowing I can contact (genetic counsellor) when I am pregnant. Knowing that I have options about tests in pregnancy"
"still anxious and waiting for results"
"found out importance of arranging marriages outside of the family so the condition does not happen again"
"I have told everyone in the family and they have all been referred for carrier testing. It has provided reassurance as no one else in the family wants an affected child it was horrible"

In the same 12-month period, there were 15 pregnancies in couples known to the service. Nine couples wanted prenatal diagnosis (PND), but the familial mutations were not known in four of these cases. Of the five couples who did have PND, two affected pregnancies were terminated and three unaffected pregnancies continued to term. Of the four couples who declined prenatal testing that would have been available, one was related to a treatable disorder, one was a late onset disorder and the other was associated with a variable phenotype. One couple had decided to defer pregnancy until prenatal diagnosis became available. For ongoing pregnancies at risk of specific genetic disorders, it was possible to put birth and neonatal care plans in place.

Educational sessions

Over a period of 18 months, eight half-day training sessions have been provided to a total of 130 health professionals and a wide range of community staff. In addition, specifically tailored training sessions were requested and provided to 39 health visitors, 28 midwives and 24 GPs. All of these sessions generated interactive debate. There was a 90 % response to the feedback questionnaires given at the end of each session. The feedback highlighted that before the sessions, primary care workers lacked confidence in assessing or discussing genetic issues, were unsure how to access genetic services and were unclear about whose responsibility it was, often assuming that someone else would have done this. As a result, a direct access referral pathway to the specialist genetic counsellor clinic was developed and disseminated. Feedback comments from participants were positive and included the following:

"I am sure we can have conversations with extended members of a particular family as it is likely that the condition could be repeated when there are close relationship marriages"
"Worthwhile project for families who have not been able to access the right type of information in the past on recessive conditions and the support and advice available through a local Genetics Service"
"I have an increased awareness of inherited disorders and feel confident in discussing that with families"
"Excellent training which was very informative and should be offered to all frontline staff working in the health professions"

Following liaison with Child Death Overview Panels, additional information on ethnicity and consanguinity is being recorded where known and the genetic counsellor is now being notified of all deaths in children of South Asian families, where there is a definite or probable genetic condition. The genetic counsellor will then recommend referral to clinic genetics for families not already known to the service.

Genetic outreach worker role

The first genetic outreach worker (Urdu speaking) was appointed in March 2014 and the second (an experienced midwife) in October 2014. Both are employed part time, working 3 and 4 days a week, respectively. Following their appointment and training, the outreach workers established support groups for parents in existing Children's Centres, providing open access for them to ask questions. Through these sessions, the outreach workers have identified families who would benefit from referral to paediatric, genetic or other support services. In addition, they have also accompanied families to hospital appointments and provided follow-up contact for families who had attended the genetic clinic and required further support. On behalf of individual families, they have additionally provided information about lay support groups, liaised with schools for children with special needs, adult social care services, carer's services, parent partnership services and social workers, as well as GPs and primary health care teams. Altogether, the outreach workers have had face to face contact with 106 families, mostly those attending the clinical genetic service.

Informal feedback from families who had contact with the outreach workers has been very positive, indicating that they valued the information they had received and the way it was given.

Feedback included comments such as

"For the first time I understand the implications for my daughter and my other children but I feel reassured that I am more aware and understand the choices I can help my children make in the future".

Discussion

The main objectives of the interventions described above were to

- Raise awareness of the availability of local genetic services
- Raise awareness of genetic issues related to consanguinity among primary health and social care workers
- Ensure primary health and social care workers have the knowledge and confidence to identify families at risk, give correct initial information and arrange referral to genetic services as appropriate
- Raise awareness of genetic issues within the South Asian community
- Provide support to families affected by autosomal recessive disorders
- Work with diverse primary health care and community services
- Build confidence in the genetic service
- Improve access to, and uptake of, genetic services for South Asian families affected by genetic disorders

The approach we took reflects the recommendations of the WHO (1985) which recognises the cultural and social importance of consanguineous marriage and that attempting to discourage this custom at population level is undesirable (Hamamy et al. 2011). The family-centred approach we have employed is likely to be particularly effective in consanguineous populations where deleterious autosomal recessive gene mutations cluster within families (Modell 1991; Darr 1997). Our clinical involvement focussed on families in which the diagnosis of an autosomal recessive disorder has been made in an individual, since this indicates that other members of the extended family may be carriers of the condition and at risk of having similarly affected children themselves if they marry blood relatives (Ahmed et al. 2002; Darr et al. 2015; Bennett et al. 2002; Modell and Darr 2002).

Our approach integrates the offer of information and support to the parents of affected children and relatives in the extended family, with a community engagement programme aimed at increasing genetic literacy in the wider community to promote accurate information and reduce stigma, which families perceive to be major problems (Khan et al. 2010; Darr et al. 2013). Marriage within families creates connections that may enable dissemination of information and provision of support. Our experience indicates that this needs to be facilitated in a culturally acceptable and empowering manner and can be difficult to achieve without active involvement and support from clinical genetic services (Khan et al. 2010).

In developing a three-stranded approach, our priority was to establish a family-centred genetic counsellor service as a prerequisite to raising awareness in the community and increasing the demand for the service from families who would benefit. This also gave us the opportunity to build trust and confidence within the local South Asian community on which to base further engagement initiatives.

Community engagement is recognised nationally and globally as key in empowering the public to engage with genetics (Department of Health 2012; Reilly 2000). Successful community education initiatives were reported by the Enhanced Genetic Service Project in Birmingham (Alberg et al. 2014), especially when interactions were face to face. However, debate around the link between cousin marriage and disability needs to be firmly supported by the provision of appropriate clinical services to empower families to make informed decisions about marriage and reproductive options.

Despite the subject of consanguinity, genetics and disability being portrayed as "sensitive" (Buxton 2008) and assumed to be an issue that the Pakistani community are reluctant to acknowledge, (Ahmad 1996) this is not our experience. Our work has shown that families are receptive and willing to engage with this topic and associated services that are provided in a sensitive and culturally appropriate manner. Greater genetic literacy in the population enables conversations in the community about consanguinity and recessive disorders. A community-based approach enables genetic information to be available to young people about consanguinity-associated genetic risk before they start their own families. This information is generally accepted if given in an appropriate manner (Khan et al. 2010; Darr et al. 2013), but it is important that this message reaches older members of the family as well, who may be important decision makers within the family (Alberg et al. 2014). It has been suggested that the trend for consanguineous marriage is decreasing (Darr and Modell 1988; Bittles 2013), but this may be largely attributable to social rather than medical influences (Darr et al. 2013).

A number of approaches to raising genetic literacy among communities that practice consanguineous marriage have been attempted in England, with different messages and communication strategies, (Salway et al. 2012; Salway et al. 2016, and Alberg et al. 2014). The impact and success of any approach is likely to depend on the demographics of the local population and whether local partners are willing to actively support this issue (Salway et al. 2012). We have had positive feedback from families engaging with the genetic service as a result of our work, comparable with our previous experience (Khan et al. 2010). Changes in cultural practices leading to improvement in health outcomes may take several years to detect, as high risk families can only be identified currently after the birth of the first affected child. It will therefore be

important to follow trends in infant and childhood mortality as well as morbidity.

As a result of our work, there is greater access to genetic counselling for consanguineous families at risk of recessive disorders in BwD. There has also been an increase in the number of referrals from midwives and GPs directly to the local genetic counselling service since 2014, indicating that when frontline staff are better informed about consanguinity-associated genetic risk and relevant services, they can identify and refer families appropriately. Having a local service and liaison with a named genetic counsellor has been a key factor in this. Families at risk are also being identified through the community engagement work and are being identified and referred by the genetic outreach worker.

In part, the success of our service development can be attributed to the strong and continuing working partnership established between the Regional Genetic Service, Public Health, Primary Care, the NHS Hospital Trust and the VCF sector. Regular updates are presented to the Local Safeguarding Children's Board, Infant Mortality Prevention Action Group and the Health and Wellbeing Board. To ensure sustainability, there must be continued engagement with key commissioners across the Local Authority, CCG and NHS England and collaborations that address not only the health needs of these families but also their social and educational needs, connecting them with community support services that empower the families to identify and address their own needs.

In conclusion, we have developed an extended genetic service addressing the needs of South Asian families who are at increased risk of autosomal recessive disorders that is sustainable and led by a specialist genetic counsellor as part of the Regional Genetic Service but in partnership with public health and with established links to other key agencies and the voluntary sector. We suggest that this may be a model that could be adopted in other regions in the UK with similar population demographics.

Acknowledgments We thank the families who have provided feedback on the service, to all the health professionals who have worked with us to develop and establish this service. Angela Allen, Chair of the Blackburn with Darwen Families and Health Wellbeing Consortium, has been a key in helping us establish and develop the role of the genetic outreach worker. Her role and support in developing the community engagement aspect of the service has been crucial

Compliance with ethical standards

Conflict of interest The authors declare that they have no conflict of interest.

Funding This study was funded by Blackburn with Darwen PCT until 31 March 2013. From 1 April 2013, Community Education and Engagement by Blackburn with Darwen Council Public Health and Genetic Counselling were funded by NHS England. There was no funding through grants.

Ethics approval This article does not contain any studies with human participants or animals performed by any of the authors. This was a clinical service development that did not need ethics approval. The service was provided through the regional genetic service.

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