JEWISH GENETIC DISORDERS

A review of awareness, education and services in the UK

December 2011

Jewish Genetic Disorders UK

JGD UK would like to thank The Andor Charitable Trust for their generous contribution to this project.

The project team would like to express their thanks to everyone who contributed to this research, including all the organisational representatives and professionals who shared their time and experience, and the reviewers for their valuable comments.

Research team

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This report has been written in good faith reflecting the information provided by participating organisations and professionals during the 2011 research period. JGD UK will be grateful for any updates and comments relating to the report content.

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About JGD UK

Jewish Genetic Disorders UK (JGD UK) is a registered charity dedicated to improving the awareness, prevention and management of Jewish genetic disorders across the Jewish, healthcare professional and wider community.

For more information visit www.jewishgeneticdisordersuk.org

Registered charity no. 1134935

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Overview

This report provides the first-ever review of awareness, education and services in the UK for Jewish genetic disorders (JGDs).

The findings and recommendations provide a platform for key stakeholders to work together to develop an effective programme of positive action to address this important issue for the Jewish community and for others in the UK.

Introduction

Over the past 30 years, research has identified a group of JGDs that include diseases that are fatal in childhood and others associated with chronic disability and premature death in adulthood.

JGDs refer to genetic conditions that, though not exclusively Jewish, have a higher prevalence among people of Jewish ancestry relative to the general population.

Although the incidence of many of the JGDs is low, it is not uncommon for people of Jewish ancestry to be 'carriers'[1] of one or more of these conditions. Carriers do not have the disorder themselves, and may have no known family history of the condition, but are still at risk of having affected children or future generations.

1 in 5 Ashkenazi Jews are carriers of at least one Jewish genetic disorder[2].

Fortunately, with the help of scientific developments, the genetic causes of these disorders have now largely been identified. Furthermore, accurate diagnostic and screening tests have been created, and some effective treatments developed.

Despite these advances, Tay-Sachs disease is still the only JGD for which carrier screening[3] is available via the NHS and for which any established community education exists in the UK. For the other disorders, some of which are more prevalent and others which are as serious as Tay-Sachs, there is no NHS-funded screening or community outreach education.

In 2010, the charity JGD UK was established to improve the understanding, prevention and management of JGDs across the Jewish, healthcare professional and wider community.

This piece of qualitative research has been carried out by JGD UK with the following objectives:

- To establish a picture of current awareness, education and services regarding JGDs in the UK.
- To provide practical recommendations for positive action to address the gaps identified.
- To provide a valuable resource to help develop future educational, referral and support initiatives that will more effectively inform and support those affected by JGDs.

Key findings

Awareness and education

Awareness of the specific Ashkenazi Jewish connection of the genetic disorders[4] covered in this research was found to be generally very low among both Jewish community and healthcare organisations. Excluding Tay-Sachs and the BRCA 1 and 2[5] associated cancers, any familiarity tended to be based on personal experience, through contact with either a family or community member affected by a particular disorder.

This is perhaps unsurprising given there is no comprehensive awareness programme running in the Jewish community, the only exception being the ultra-orthodox community, which has a long-running pre-marriage screening programme. In addition, formal training for healthcare professionals, apart from genetic counsellors, does not make any specific reference to JGDs.

Patient group representatives were aware of the Jewish dimension of the specific JGD they covered. However, most indicated that, in their experience, awareness was unlikely to extend beyond professionals working with the conditions and affected families.

Services

Tay-Sachs disease continues to be the only JGD for which the National Screening Committee has formally approved carrier screening for the Ashkenazi Jewish population in the UK.

In terms of other NHS services, the more common conditions tend to be well supported by specialist provision, whereas the very rare disorders have little in the way of dedicated specialist care. For many of the JGDs, obtaining a clear diagnosis is often a difficult and lengthy process, and specialist expertise and support in the UK is limited.

Most of the individual disorders have some patient group representation in the UK. These groups vary greatly in terms of their service range and focus depending on their size.

International

International comparison with the USA, Canada, Israel and Australia shows that whilst community education and screening initiatives for JGDs may vary, all these countries are significantly more active in addressing the issue of JGDs than in the UK.

Recommendations

Analysis of the research identified four key measures that need to be put in place in the UK:

- A targeted programme of awareness-raising and education
- A more comprehensive screening programme
- An effective information and signposting system
- A regular review process

Building on this consultative project, we now have a real opportunity to work collaboratively to capitalise on scientific advances and make a significant difference in the prevention and management of these genetic conditions.

At a time when medical advances have enabled us to identify a Jewish connection to genetic disorders and have established ways of assessing and addressing them, our collective goal should be for no Jewish parent in the UK to ever again ask, "How could we not have known?"

Methodology

This piece of qualitative research was carried out over a one-year period by a small research team, with guidance from members of the JGD UK's Board of Trustees and its Science and Medical Advisory Committee.

Disorders covered

The research focused on genetic disorders that have a higher prevalence among Ashkenazi Jews relative to the general population, given that approximately 90–95% of British Jewry is of Ashkenazi ancestry.

Listed below are the 12 conditions included in the research. Further information about the disorders is summarised on pages 18–19 of this report. For the purposes of this report, the term 'Jewish genetic disorders (JGDs)' will refer only to these conditions.

Bloom syndrome

Gaucher disease (type 1)

Breast and ovarian cancer (BRCA 1 & 2)

Generalised dystonia (DYT1)

Canavan disease

Mucolipidosis IV

Cystic fibrosis

Niemann-Pick disease (type A)

Familial dysautonomia

Tay-Sachs disease

Fanconi anaemia (type C)

These disorders, which are currently covered by JGD UK, were selected largely on the basis of their carrier frequency[6] and severity.

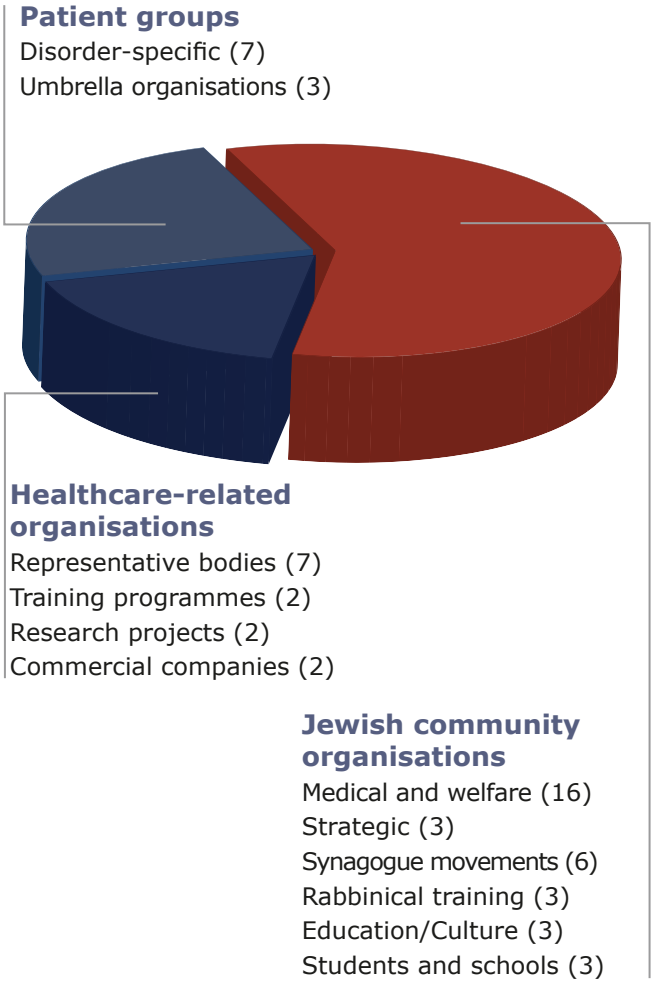
It is important to note that cystic fibrosis and the cancers have certain mutations[7] that are more prevalent among Ashkenazi Jews even though the disorders themselves may not be more common than in the general population.

Other JGDs have been identified, including factor XI deficiency, non-classical adrenal hyperplasia and glycogen storage disease (type 1a), as well as conditions more common in non-Ashkenazi Jews. JGD UK hopes to cover more conditions in the future.

Participants

A broad range of organisations and professionals with a clear actual or potential interface with JGDs were identified. **Over 60 organisations** from the Jewish, healthcare professional, and patient group communities were included in the research. A list of participant organisations can be found on page 16 of this report.

The organisations included are by no means exhaustive. During the research, some additional contacts were identified that JGD UK will be in touch with as part of future research and outreach work.



Data collection and analysis

Information was gathered from key representatives from the central offices of the participant organisations and from a small sample of individual healthcare professionals working in communities with a large proportion of Jewish individuals.

A variety of methods were used for data collection, including: e-mail, telephone interviews, face-to-face interviews, on-line searches and research review.

International experience relating to JGDs screening and education was also briefly reviewed to serve as a point of comparison and to explore learning opportunities for the UK.

The research findings were analysed to identify key issues and challenges; and recommendations for positive action were made.

Main findings

Awareness and education

The Jewish community

- **Research across Jewish community organisations in the UK strongly suggests that awareness of Jewish genetic disorders (JGDs) in general is very low.**

- Across the many sectors of UK Jewry, only the ultra-orthodox community was found to be well aware of JGDs, attributed mainly to the work of the screening organisation Dor Yeshorim[8].

- Only a very small proportion of the organisations contacted had an awareness of a group of genetic disorders that are relatively more common among Jewish people. These were Chana, the Jewish Medical Association (UK) and the Board of Deputies of British Jews.

- Levels of awareness were found to vary across the individual disorders:

  - **Tay-Sachs** appeared to be well known as a genetic condition that affects Jewish people, largely as a consequence of Jewish Care's screening and awareness-raising.

  - Findings indicated a reasonable and growing awareness of the increased risk among Ashkenazi Jews of developing **breast or ovarian cancer** associated with BRCA 1 and 2 mutations. Chai Cancer Care is very familiar with the Jewish aspect of certain cancers. In addition, the GCaPPs[9] (Genetic Cancer Prediction through Population Screening) study has also raised the profile of BRCA 1 and 2 mutations across the Jewish community.

  - There was some familiarity with **Gaucher disease (type 1)**, attributed in part to its relatively high prevalence as compared to most other JGDs, and also due to the work of the well-established Gauchers Association.

  - Some small awareness of **familial dysautonomia** was indicated, helped by the awareness-raising efforts of the Dysautonomia Society of Great Britain and other affected families, all of which are Jewish.

  - For **Bloom's syndrome, Canavan disease, Fanconi anaemia (type C), generalised dystonia (DYT1), mucolipidosis IV and Niemann-Pick disease (type A)** – half of all the disorders researched – awareness was found to be negligible.

- **There is no systematic programme of awareness-raising or information dissemination about JGDs in the Jewish community.**

- **Across the wide range of organisations contacted, an interest in JGD-related issues was expressed together with broad support for working collaboratively with JGD UK on awareness-raising initiatives.**

Healthcare professionals

- **Awareness of JGDs among healthcare professional groups reviewed appears to be generally low, though there was a reasonable awareness of Tay-Sachs disease and the higher prevalence of certain BRCA mutations among people of Jewish ancestry.**

- Healthcare professionals tended only to be aware of the Jewish aspect of the other conditions if they had actual experience of an affected patient, or were specialists in an area that included one or more of the conditions, e.g. metabolic paediatricians.

- With the exception of genetic counselling, none of the healthcare professionals' training curricula reviewed made any specific reference to a group of genetic disorders that are relatively more common among Jewish people.

- GP training covers a general understanding of genetics but does not refer specifically to JGDs or mention Tay-Sachs screening despite this being approved by the National Screening Committee.

- Genetic disorders and inheritance in general are covered in the training curricula for obstetricians and gynaecologists, with Tay-Sachs being given as an example.

- The general paediatric curriculum covers diagnosis of genetic and metabolic conditions, including some disorders covered in this report. However, the Jewish aspect is not mentioned.

- Clinical geneticists' training focuses on acquiring skills to research specific conditions as they arise in clinical practice. As such they are taught in general about ethnic differences in the incidence of genetic disease and about the purpose of screening, but JGDs are not mentioned specifically in the curricula.

- Neither midwife nor health visitor training covers the subject of JGDs as standard. Some awareness was indicated among midwives working in areas with sizeable Jewish populations; however, screening is not offered routinely or consistently.

- The recently launched Jewish health section of the Healthtalkonline[9] website captures personal experiences of some JGDs as an awareness-raising tool for healthcare professionals and people affected.

- **There was a clear sense that it is neither practical nor necessary for all these healthcare professional groups to be taught about all JGDs – especially the rare ones. Yet, positive interest was expressed to learn more about the concept in general, and screening opportunities, as long as the information was targeted, accessible and sensitive to time pressures.**

Patient groups

- The majority of JGDs have some patient group representation in the UK; however, most of the groups cover a particular JGD as only a small part of their support for a wider family of genetic conditions.

- **Bloom syndrome** and **Canavan disease** have no specific representation in the UK.

- Jewish Care and the Dysautonomia Society of Great Britain are Jewish-community-based organisations that focus exclusively on **Tay-Sachs** and **familial dysautonomia** respectively. Both groups are very aware of the Jewish aspect of the disorders they focus on and are involved in various activities to raise awareness of these disorders in the community.

- Every representative contacted was familiar with the issue of higher Jewish prevalence regarding the specific JGD that they covered, and a few of the groups included a line about this on their website or, relevant information leaflets.

- Most patient group representatives indicated that, in their experience, awareness of the higher relative Jewish prevalence regarding the different JGDs was likely to be limited to affected families and professionals working with the conditions.

- Each patient group was involved in various generic awareness-raising activities for the range of conditions that they covered but none of these were JGD-specific.

- **All the patient groups welcomed the opportunity for an ongoing dialogue with JGD UK to support awareness-raising initiatives of mutual interest.**

Services

Genetic screening and testing[11]

- **Tay-Sachs disease is the only JGD for which the National Screening Committee has formally approved carrier screening for the Ashkenazi Jewish population in the UK.**

- Although the Tay-Sachs screening service has reduced disorder incidence in the Ashkenazi Jewish population, a 2009 report[12] by the PHG Foundation noted that there is no routine or readily available information about the service, its activity or its outcomes. The report made strong recommendations for screening service improvement which, to date, have not been taken forward.

- Privately funded carrier testing for a panel of recessive[13] JGDs is available to the ultra-orthodox community via the screening organisation Dor Yeshorim, and to the wider population via a few of commercial companies.

- **Guy's and St Thomas' NHS Foundation Trust and Great Ormond Street Hospital are each in the process of setting up a genetic carrier testing service for a panel of recessive JGDs in response to dialogue with JGD UK. This service will be offered privately and delivered within an NHS context with associated counselling.**

- The NHS does cover genetic testing for individual JGDs in cases where there is an established family history, or known carriers in the family, of a particular disorder – this testing can be accessed via a GP referral to a Regional Genetics Service.

- According to NICE guidelines, women classed as having an 'increased risk' of breast or ovarian cancer given their family history may be offered genetic testing for BRCA 1 and 2. These guidelines include separate categories regarding family history assessment and BRCA testing criteria for families of Jewish descent.

- Pre-implantation genetic diagnosis[14] for couples with a high risk of passing on a particular JGD is now available at a number of Human Fertility and Embryology Authority licensed centres. NHS funding may be available to some couples, depending on whether they fit primary care trust criteria.

- Prenatal genetic diagnosis[15] is also available to couples at increased risk via specialist fetal medical units with the support and co-ordination of clinical genetics departments.

Treatment and management

- Services for the different JGDs are organised according to medical specialities.

  The more common conditions tend to be well supported by specialist services, whereas the very rare disorders have little in the way of dedicated specialist care.

  For the less common JGDs, getting a clear diagnosis is often a difficult and lengthy process and specialist expertise and support in the UK is limited.

  Some transitional care issues were raised in relation to patients affected by disorders whose management moves from paediatric to adult services.

- Paediatric metabolic services diagnose and manage the care of children affected by the following metabolic JGDs: **Tay-Sachs disease, Niemann-Pick disease, Mucolipidosis IV and Gaucher disease.**

- In addition, **Gaucher disease** is well supported by 7 lysosomal storage disorder specialist units in the UK. These units, staffed by multi-disciplinary teams, offer a wide range of services to help diagnose, treat (including provision of enzyme replacement therapy) and manage the care of child and/or adult Gaucher disease patients.

- Across the UK, there are 24 paediatric and 24 adult specialist **cystic fibrosis** centres that deal with the diagnosis of, manage the care of, and help monitor affected individuals.

- Extensive and well-established cancer services exist in the UK for **breast and ovarian cancer** patients and for people found to carry the BRCA mutations that put them at 'increased risk' of developing these cancers.

- The treatment and care of **generalised dystonia** patients is managed by neurology departments, usually via their movement disorder clinics. A variety of treatment options are now available that can either

be offered by neurologists directly or through referral to related specialist departments, e.g. neurosurgery.

- **Fanconi anaemia** patients tend to be managed by their local haematologists. There is also a UK Fanconi anaemia clinical network made up of clinicians who have expertise or an interest in this disorder. They share best practice and decide on treatment standards and protocols.

- Various specialists, e.g. in the areas of neurology, respiratory, musculo-skeletal and genetics, are involved with managing the different medical needs of **familial dysautonomia** patients; however, affected families sometimes lack the support of a dedicated clinician responsible for co-ordinating the multidisciplinary expertise needed.

- In the UK, there are no dedicated specialists, treatment centres or guidelines for **Canavan disease** or **Bloom syndrome** management. Canavan patients tend to managed by local paediatric neurologists, with input from metabolic and genetics services. Bloom syndrome patients are usually cared for by local haematology departments.

Additional support services

- Patient groups vary greatly in terms of their service range and focus; larger groups generally represent the more prevalent conditions and offer a broad spectrum of support, whereas smaller groups tend to offer fewer services which focus on the key identified needs of their members.

- **Bloom syndrome** and **Canavan disease** have no dedicated patient groups in the UK; however, some information and support for these conditions can be accessed via the umbrella groups Contact a Family and Climb. Also, Genetics Alliance offers some representation for these conditions, as it does for all rare genetic disorders.

- **JGD UK is the only organisation in the UK that focuses exclusively on JGDs. It aims to work collaboratively with other organisations to raise awareness and to facilitate access to the best available information, services and support relating to JGDs.** Funding from Jeans for Genes has been awarded to develop a bespoke information and referral service for families with affected children and young people.

International experience

- **Community education and screening initiatives for JGDs vary across the USA, Canada, Israel and Australia. However, all these countries were significantly more active in addressing the issue of JGDs than the UK.**

- National guidelines recommending carrier screening for JGDs among the Jewish population exist in the USA, Canada and Israel. Disorders covered by the guidelines vary; however, all include Tay-Sachs disease, familial dysautonomia and Canavan disease as a minimum.

- In the USA, a number of community-based organisations exist that are dedicated to raising awareness and promoting screening for JGDs, and in many cases helping to subsidise screening which otherwise would need to be privately funded.

- Although the countries reviewed differ culturally and in terms of their medical systems, the UK can still learn from their outreach community education experience and approaches to facilitating screening for an expanded panel of disorders.

Recommendations

ONE: DEVELOP AN EFFECTIVE PROGRAMME OF AWARENESS-RAISING AND EDUCATION

To promote better understanding of Jewish genetic disorders across the Jewish community and among health professionals, organisations and individuals.

For the Jewish community

A systematic and targeted education programme urgently needs to be developed and delivered in partnership with the main Jewish community organisations. It is essential that the community is better informed about JGDs in order to help reduce the incidence and impact of these conditions.

Tailored packages for the different types of organisations and movements may be necessary, supported by a network of champions/ambassadors within the different grass-roots organisations. Halachic considerations also need to be taken into account by directing community members to their respective rabbinical authorities for guidance.

With regard to non-affiliated members of the community, it will be necessary to generate some coverage in the general media.

For healthcare professionals

An effective communications programme needs to be created for healthcare services particularly focused on areas of concentrated Jewish population. This should include education about the disorders and their specific Jewish prevalence, as well as effective signposting to services and patient groups.

Information materials and dissemination channels need to be developed in consultation with working groups of healthcare professionals to help ensure positive impact. The focus initially should be on GPs as primary gatekeepers to other services; however, outreach education should also extend to other healthcare professionals such as obstetricians and gynaecologists, paediatricians and midwives.

TWO: ESTABLISH A MORE COMPREHENSIVE JGD SCREENING PROGRAMME

Expand screening for JGDs

Tay-Sachs screening should be expanded to include a broader panel of recessive JGDs to help reduce disorder incidence and enable those at risk to have available the widest reproductive options possible. The screening should be targeted appropriately in the Jewish community, designed and delivered in a way that ensures that those accessing the services are responsibly informed and supported.

The inclusion of disorders in the panel should be based on severity, carrier frequency and test accuracy. It would not be appropriate to include testing for BRCA and dystonia mutations because these are dominant[16] and require more complex counselling. There should be a review every two years to assess whether or not the panel needs amending.

As, in the immediate future, expanded screening options are likely to need to be self-funded it is important that service costs are reasonable and financially accessible to all at risk. Additional resources will be needed to help subsidise screening for low-income groups and achieve effective levels of service uptake.

It is advised that a health economist carry out a cost-benefit analysis of expanded JGD screening in the UK as a platform for dialogue with policy makers about possible inclusion of the service within the NHS.

Consolidate existing Tay-Sachs screening

Efforts need to be made to help promote implementation of the important recommendations made by the 2009 PHG foundation report. In particular, the programme needs to be maintained and formally constituted by the NHS, with improvements being made to the availability of information regarding the service, its activities and outcomes.

THREE:
DEVELOP AN EFFECTIVE INFORMATION AND SIGNPOSTING SYSTEM

Create a centralised information and referral resource

A centralised resource is needed to improve access to information and enable effective signposting to available expertise and other resources. This is particularly essential to help those with a new diagnosis, or struggling to obtain a clear diagnosis, to access the best available information and support in a timely manner.

Consolidate collaborative relations

There is a need to consolidate relations between JGD UK, relevant patient groups and Jewish community welfare and other organisations established through this research, to enable better flow of information and more effective referral. In addition, working collaboratively will create a stronger collective voice for issues of mutual interest.

FOUR:
DESIGN AND IMPLEMENT AN EFFECTIVE REVIEW PROCESS

Introduce a review process to capture and assess the impact of relevant initiatives on awareness levels, which would also help identify gaps and needs. This may include surveying representative samples of the Jewish and medical communities; measuring JGD UK website traffic; monitoring screening uptake; and evaluating pre and post surveys of awareness levels in relation to education outreach.

When local healthcare and social service providers carry out mapping of community needs in areas of Jewish population density, information dissemination and screening regarding genetic issues should be included.

Given raised awareness levels of JGDs following a successful educational campaign, it will also be necessary to review access to, and uptake of, existing services with a demand-led focus in relation to possible modifications or new provision.

Notes

1. **Carrier:** A person who has one altered (mutated) copy of a gene and one normal copy of the same gene. For recessive disorders (see note 13 below), carriers are unaffected by the disorder but are at risk of passing it on to their children.

2. **Gross, S.J., Pletcher, B.A. and Monaghan, K.G. (2008).** Carrier screening in individuals of Ashkenazi Jewish descent. *Genetics IN Medicine,* 10, 54–56.

3. **Genetic carrier screening:** A process of systematically offering a carrier test to a whole population (or sub-population) of healthy people, for a gene mutation associated with a particular genetic disorder.

4. **Genetic disorder:** A condition resulting from an alteration, or mutation, in one or both copies of a specific gene, leading to the development of symptoms.

5. **BRCA:** BRCA 1 and BRCA 2 are tumour suppressor genes that regulate cell growth. Individuals that have certain mutations on these genes are predisposed to a higher risk of developing cancer, particularly breast and ovarian cancer. In addition, men are at increased risk of prostate cancer; the precise levels of risk are still being determined.

6. **Carrier frequency:** A number that describes how many people in a population are carriers (have one altered copy of a gene and one normal copy of the same gene) for a genetic disorder.

7. **Mutation:** A structural change in a gene. It can be an alteration to a gene's size, arrangement or molecular sequence.

8. **Dor Yeshorim:** An international genetic screening organisation established within the Orthodox Jewish community to prevent JGDs. They offer testing for a panel of recessive disorders to unmarried, previously untested individuals who intend to find a spouse by arranged marriage. Results are not supplied but a PIN number is given and checked against that of a potential spouse to assess compatibility.

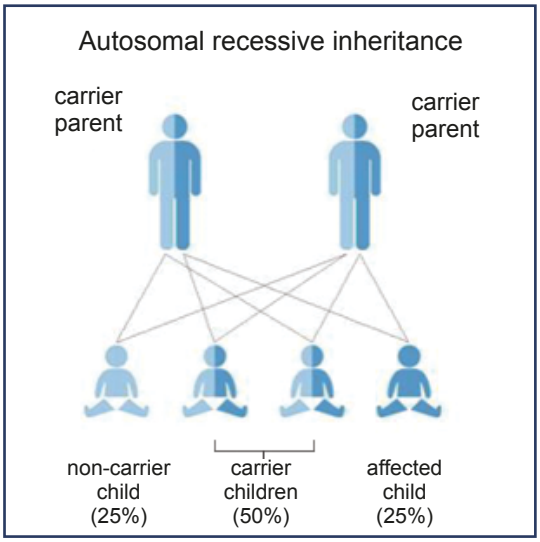
9. **Genetic Cancer Prediction through Population Screening (GCaPPs):** A UCL-based research study evaluating the feasibility of offering everyone over 18 in the Ashkenazi Jewish population a genetic test to identify those at increased risk of developing certain cancers, or passing these on to their children.

10. **Healthtalkonline:** An award winning website that covers over 50 medical conditions and allows people to share in more than 2000 personal accounts of health and illness. The information on the website is based on qualitative research, led by a team of senior researchers based at the University of Oxford.

11. **Genetic testing:** The testing process used to detect the presence or absence of, or a change in, a particular gene or chromosome.

12. **Burton, H., Levene, S., Alberg, C. and Stewart, A. (2009).** Tay Sachs Disease carrier screening in the Ashkenazi Jewish population: A needs assessment and review of current services. Published by PHG Foundation.

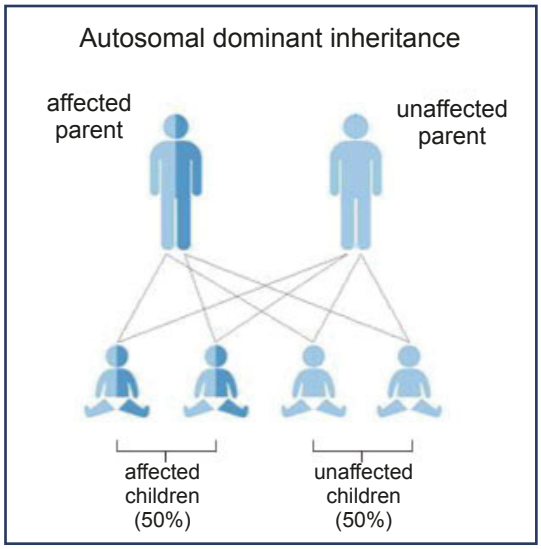
13. **Recessive:** A recessive disorder is one that results only when an individual inherits two copies of a specific disorder-related mutation, one from each parent. With recessive conditions a single fully functional copy of a gene is sufficient to compensate for the mutated copy, preventing the disorder developing. Where both partners of a couple are carriers for the same disorder gene, there is a 1 in 4 chance in each pregnancy for the child to be affected.



14. **Pre-implantation genetic diagnosis:** Use of genetic testing on one or two cells taken from a live early-stage embryo created by IVF. It is usually carried out in order to determine whether the embryo is affected by a serious genetic disease. Unaffected embryos would be implanted in the uterus.

15. **Prenatal genetic diagnosis,** also known as prenatal testing, refers to the test that a pregnant woman would have to determine whether the baby she is carrying is affected by, or at risk of being affected by, a genetic condition.

16. **Dominant:** A disorder is dominant when an alteration in just one copy of a pair of genes is sufficient to cause a detectable trait. Individuals who have one mutated copy of a gene for a dominant disorder are likely to be affected. Where one partner of a couple has an abnormal copy of a gene for a dominant disorder, there is a 1 in 2 chance in each pregnancy for the child to inherit the abnormal gene and potentially be affected by the disorder.



Participant organisations

Key representatives of the organisations below contributed to this research.

Jewish community organisations

Medical and welfare:

Birmingham Jewish Community Care
Camp Simcha
Chai Cancer Care
Chana
Dor Yeshorim
Federation of Jewish Services
Gateshead Jewish Family Care Services
Jewish Care
Jewish Care Scotland
Jewish Child's Day
Jewish Marriage Council
Jewish Medical Association UK
League of Jewish Women
Leeds Jewish Welfare Board
Merseyside Jewish Community Care
Norwood

Strategic:

Board of Deputies of British Jews
Institute for Jewish Policy Research
Jewish Leadership Council

Synagogue movements:

Assembly of Masorti Synagogues
Federation of Synagogues
Liberal Judaism
Movement for Reform Judaism
Spanish and Portuguese Jews' Congregation
United Synagogue

Rabbinical training:

Leo Baeck Rabbinical College
Masorti Rabbinic Team Training
Peir – United Synagogue Professional Development

Education/Culture:

Limmud
Jewish Community Centre for London
London Jewish Cultural Centre

Students and schools:

UJS Hillel
Jewish Free School, London
King David High School, Manchester

Patient groups

Disorder-specific groups:
Cystic Fibrosis Trust
Dysautonomia Society of Great Britain
Fanconi Hope
Gauchers Association
Niemann-Pick Disease Group (UK)
Society for Mucopolysaccharide Diseases (MPS)
The Dystonia Society

Umbrella organisations:
CLIMB
Contact a Family
Genetic Alliance UK

Healthcare-related organisations

Representative bodies:
Joint Royal Colleges of Physicians Training Board (Clinical Genetics)
Nursing and Midwifery Council
Royal College of General Practitioners
Royal College of Nursing
Royal College of Obstetricians and Gynaecologists
Royal College of Paediatrics and Child Health
Specialist Commissioning Advisory Group, London

Training programmes:
Cardiff University (MSc Genetic Counselling)
Manchester University (MSc Genetic Counselling)

Research projects:
GCaPPs
Healthtalkonline (Jewish health project)

Commercial companies:
The Doctor's Laboratory (TDL)
This is my: Health Screening and Ultrasound Centre

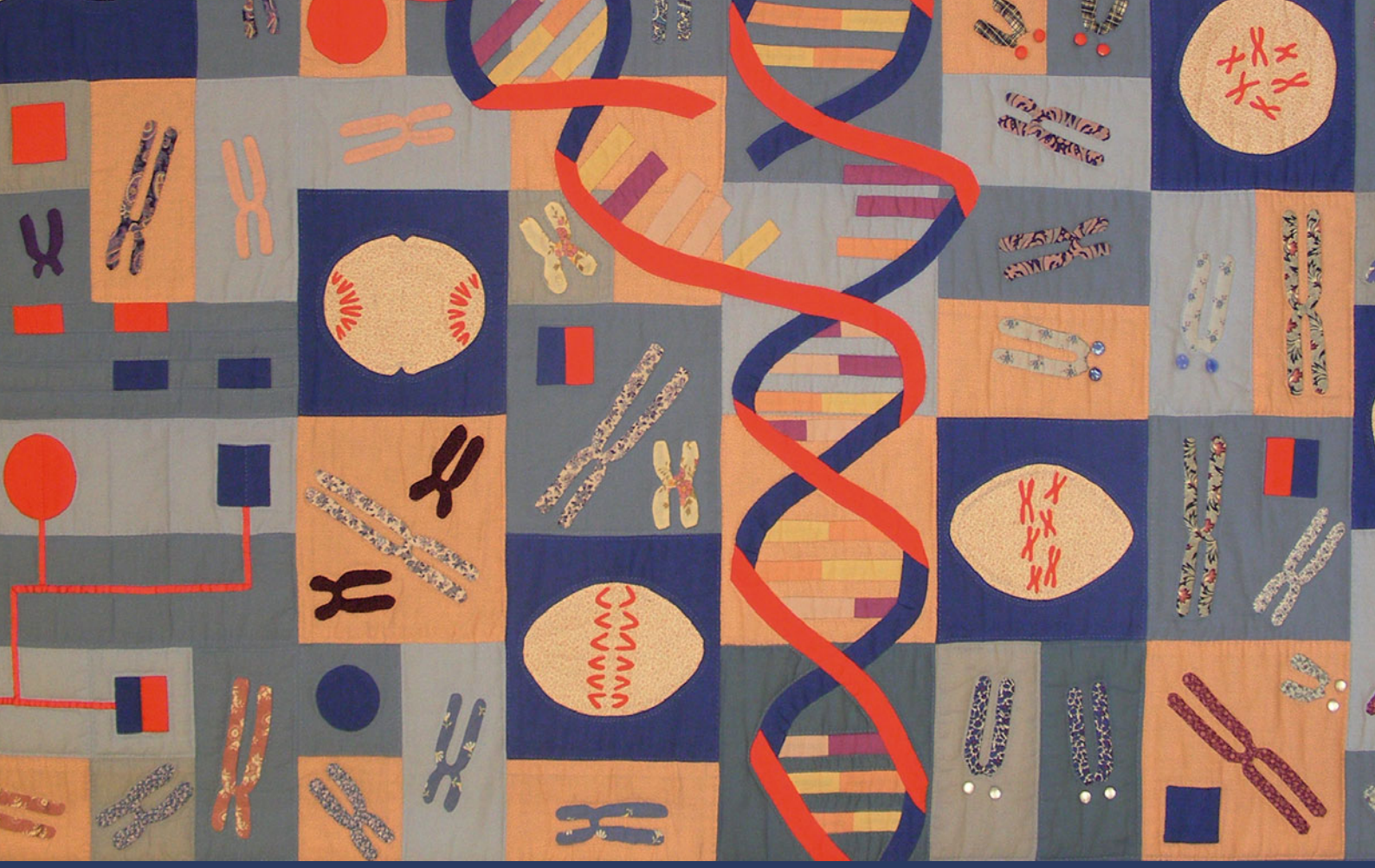
Summary table of genetic disorders

The table below presents key information about the genetic disorders included in this research.

Disorder	Description	Mode of inheritance	Carrier frequency in the Ashkenazi Jewish population
Bloom syndrome	A condition characterised by poor growth, increased skin sensitivity to sunlight, infections and a predisposition to cancer. Shortened lifespan – usually cancer related before the age of 30.	Recessive	1 in 100
Breast and ovarian cancer (BRCA 1 and 2)	A disease in which cells grow and reproduce themselves abnormally, causing cancerous tumours (in the breast area for breast cancer and in one or both ovaries for ovarian cancer). Disease prognosis varies – it is life-threatening; however, in some people, treatment can effectively remove the cancer and stop disease progression.	Dominant (with variable penetrance)	BRCA 1/2 mutation carrier frequency is: 1 in 40
Canavan disease	A disorder where affected children show normal early development but then experience progressive neurological deterioration of physical and mental capabilities. Shortened lifespan (typically fatal from early childhood through to late teens).	Recessive	1 in 40–57
Cystic fibrosis*	A progressive disorder that causes the body to produce thick, sticky mucus in the lungs and digestive system. Symptoms vary but may include frequent respiratory infections, poor weight gain and progressive lung damage. Shortened lifespan (typically fatal around the age of 30).	Recessive	1 in 25–29
Familial dysautonomia	A progressive disorder that causes the autonomic and sensory nervous systems to malfunction affecting a range of bodily functions including temperature and blood pressure regulation, swallowing, tear production, pain sensitivity and response to stress. Shortened lifespan (50% live to 40 years).	Recessive	1 in 30
Generalised dystonia (DYT1)	A neurological movement disorder characterised by sustained and involuntary muscle contractions or muscle spasms. It usually has an early onset (children or young adults), with abnormal muscle contractions beginning in the lower or upper limbs and then often spreading to other parts of the body, causing physical disability, often with associated pain. Symptom severity varies and treatments are available. Lifespan is not affected.	Dominant (with reduced penetrance)	1 in 1000–3000

Disorder	Description	Mode of inheritance	Carrier frequency in the Ashkenazi Jewish population
Fanconi anaemia (type C)	A disorder characterised by a reduced production of all types of blood cells and an increased risk of cancer. Symptoms include birth defects, short stature, skin discolouration, fatigue, hearing loss and reduced fertility. Shortened lifespan (typically fatal by the age of 30).	Recessive	1 in 89
Gaucher disease (type 1)	Lysosomal storage disorder caused by an enzyme deficiency that can result in anaemia, low platelet count, easy bruising and bleeding, some form of bone disease and enlarged liver/spleen. Range and severity of symptoms varies greatly and can be treated. Lifespan tends to be unaffected.	Recessive	1 in 10–15
Mucolipidosis IV	A progressive disease affecting the brain and nervous system and characterised by significant delays in motor and cognitive development, low muscle tone and progressive visual problems. Mild to severe progressive developmental delay – shortened lifespan (ranging from 1 to 45 years).	Recessive	1 in 100–125
Niemann-Pick disease (type A)	A progressive disorder caused by a deficiency of an enzyme responsible for breaking down a specific fat in the body. Fat accumulates in various organs, brain and nervous system, causing rapid neurological decline. Shortened lifespan (typically fatal from 2 to 5 years of age).	Recessive	1 in 90
Tay-Sachs disease	A neurodegenerative disorder characterised by normal birth and development until 3–6 months then rapid and progressive brain and nervous system deterioration. Classic infantile form has a shortened lifespan (typically fatal by age 6).	Recessive	1 in 26 - 30

* Cystic fibrosis has certain mutations that are more prevalent among Ashkenazi Jews, however, the disorder itself is no more common than in the general population.



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