It's in your GENES

With Ashkenazi Jews particularly prone to certain genetic disorders, Alexandra Roach looks at the implications and how the community is responding.

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DID you know the average person walking around on the street is carrying nine genetic disorders? Yes, as is often reported, Ashkenazi Jews are more prone to certain specific conditions – such as Tay-Sachs, Canavan disease, cystic fibrosis, familial dysautonomia and Fanconi anaemia – than other ethnic groups.

But are they more likely generally to be afflicted by degenerative conditions than the rest of the population?

According to Dr Leslie Burnett, a clinical professor of genetic medicine at the University of Sydney, the answer is no.

So why do Jews with an Eastern European background grab the attention?

"The Jewish community has volunteered more than other groups and has always been very generous with genetic samples, so we have good records of what to look for when it comes to genetic conditions," the doctor tells *The AJN*.

An Ashkenazi Jew himself, Burnett was instrumental in establishing a screening program which has seen a dramatic drop in the number of Jewish children born with genetic conditions.

IT was when Burnett and his wife were expecting their second child in 1987 that they discovered they were both genetic carriers of Tay-Sachs.

Carried by approximately 1 in 25 Ashkenazi Jews, the condition usually results in death in infancy. A child affected by Tay-Sachs will most often appear to be developing normally until six months of age, when the child's motor skills, hearing and sight begin to deteriorate, usually at a rapid rate.

"When we discovered we were both carriers, my wife and I were deeply concerned our child was at risk," Burnett recalls.

"We had no way of knowing. It was a horrible feeling."

When their child was born healthy and unaffected by Tay-Sachs, the couple breathed a sigh of relief.

But Burnett knew something had to be done.

"I went and researched options available for genetic testing at the time, and how a program for Sydney could be organised," he says.

It was a long process. Burnett says that labs in Brisbane and Adelaide had the capability to test for Tay-Sachs but no such facilities existed in either Sydney or Melbourne, where Australia's largest Jewish communities reside.

Beginning in 1992, Burnett was able to offer testing in Sydney at Westmead Hospital. Patients were referred by doctors but the percentage of Jews being screened remained lower than he had hoped.

"Professor Burnett realised that to achieve the required take-up it needed to be 'owned' by the Jewish community," Wolper Hospital CEO Harry Aizenberg tells *The AJN*.

"In 1995, Professor Burnett approached the JCA to assist with a community-screening program for Tay-Sachs disease.

"The JCA then acted as marriage brokers and introduced him to Wolper Jewish Hospital."

The partnership with Wolper continues to this day and has had a phenomenal impact.

"Since the program began in 1995, not a single Tay-Sachs-affected child has been born to parents who have been tested as part of the program," Burnett says.

"Parents have been able to avoid the trauma of having a child affected by Tay-Sachs," Burnett says.

"The program has made a difference. It's an amazing result."

THE Wolper initiative was the first community-based genetic screening program in Australia and Burnett's vision of the community 'owning' it has come to pass: Jews from the most Orthodox to the most secular are happy participants.

"All sectors of the community have been extremely generous and very supportive," Burnett says.

The program provides a wide variety of services relating to genetic conditions, including information sessions and genetic counselling, all free of charge and on an anonymous basis.

Testing is offered for Tay-Sachs, cystic fibrosis, Canavan disease, familial dysautonomia and Fanconi anaemia.

The screening originally had to be done via a blood test, although advances in medical technology in recent years have allowed for testing to now be conducted with a saliva sample.

"We have education sessions for all students who attend Jewish day schools in Sydney," Burnett explains.

"We hold these sessions for, and give information packs to, students in year 11.

"They're old enough to handle the information but not distracted by their final exams," Burnett says.

"It's fine if they don't want to be tested at all or at that moment and prefer to come back at a later date.

"We tell them how they can access testing when they choose, including at Royal North Shore Hospital (RNSH) and Wolper."

Burnett says the take-up of the testing program among secondary students is 96-97 per cent.

"The thought of having a blood test with a needle used to turn some people off.

"The saliva testing, which is completely painless and requires nothing more than a mouth swab, has certainly helped to increase the take-up," he says.

The program also works with the ultra-Orthodox community, with rabbis and ethicists on hand to help navigate moral issues.

For a child to be born affected by conditions including Tay-Sachs and Canavan, both parents must be genetic carriers of the condition. Carriers are not affected by the condition themselves, and a couple who are both carriers have a 25 per cent chance of having an affected child. There is a 50 per cent chance the child will be an unaffected carrier of the condition and a 25 per cent chance of having a child who is neither affected nor a carrier.

"In the ultra-Orthodox community, the matchmakers will not introduce a couple if they are both carriers, which prevents the possibility of them conceiving a Tay-Sachs-affected child."

While Burnett stresses it is best for people to be screened well before they think about having children, the program does provide prenatal testing.

"A foetus can be tested in utero via chorionic villus sampling (CVS), which allows us to know if the foetus is affected by, or a carrier of, these conditions," Burnett says.

"Individual ovum and sperm can also be tested.

"It's about ensuring people are informed and have information early. Then they have a full range of choices available to them.

"We have specially trained genetic counsellors on hand to answer questions and discuss options, which people find particularly helpful.

"It can be a lot to take in."

LEAPS forward in technology have been significant in terms of genetic testing since the program began.

Another recent breakthrough will revolutionise the program later this year: a new, state-of-the-art gene sequencer will begin operation at RNSH and will allow for screening for dozens of conditions prevalent in Ashkenazi communities all in one test.

Aizenberg says media coverage of genetic testing which allows people to know if a genetic condition will affect their own health in later life has made some people nervous about being tested.

"It's important to remember that we are testing for your carrier status of certain mutations that have no bearing whatsoever on your health," Aizenberg says.

"It is the health of your future children that may be affected.

"The media is awash with stories of genetic testing for mutations that are predictive of your own health, but our testing program is for diseases that can only be genetically inherited when the mutation is passed on by the both the mother and father.

"It's about preparing for the future.

"After all, forewarned is forearmed."

For more information about screening, please call Wolper Hospital on (02) 9328 6077.

The conditions

TAY-SACHS, Canavan disease and familial dysautonomia all affect the central nervous system, with sufferers afflicted by an inability to control their limbs or stress response, or swallow or speak.

Cystic fibrosis causes a thick mucus to build up in the digestive tract and lungs, although it does not affect intelligence or brain function.

Fanconi anaemia interferes with the proteins responsible for repairing DNA.

Testing in Victoria

IN Melbourne, community genetic screening sessions are held twice a year at the Beth Weizmann Community Centre.

Year 11 students at Jewish schools, and year 11 and 12 students at non-Jewish schools, are provided with testing and genetic counselling free of charge.

The Melbourne genetic screening program, the Tay-Sachs Disease and Related Conditions Screening Program, is funded by the Pratt Foundation, the Gandel Foundation, the Finkel Foundation, the Besen Foundation and anonymous donors.

Testing in Melbourne is available year-round via the Austin Health Clinical Genetics Service. For further information, call (03) 9496 3027.