ELECTRONIC LETTER

A genetic screening programme for Tay-Sachs disease and cystic fibrosis for Australian Jewish high school students

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Australia has a Jewish population of about 90 000, mostly living in metropolitan Sydney or Melbourne and most are of Ashkenazi (northern and central Europe) Jewish origin.¹ While community genetic carrier testing programmes for Tay-Sachs disease (TSD) have been established since 1970 and are now operating in various forms in 15 countries,²⁻⁴ before 1993 in Australia all TSD laboratory testing was only available through a medical consultation service. Following a two year pilot study,¹ the Tay-Sachs Disease Programme (TSDP), organised by the Australasian Community Genetics Programme (ACGP), was established in 1995.

Knowledge of genetic carrier status allows people an accurate assessment of their risks for having children with conditions such as TSD, enabling at risk couples the opportunity to explore their reproductive options, which may include prenatal testing, adoption, sperm or egg donation, and more recently preimplantation genetic diagnosis.²⁻⁵ While decision making in this area is optimally made before pregnancy, there are enormous difficulties in informing those of reproductive age of the availability of genetic carrier testing and its relevance.⁵

The ideal age for population screening for autosomal recessive diseases (such as TSD) is therefore early in adulthood, when young people can make mature decisions about testing⁶ based on information provided in a forum that enables discussion and debate. The high school environment provides that opportunity and it has been successfully shown in Montreal by Mitchell *et al*⁷ that screening for genetic carriers for TSD (1973-1992) and β -thalassaemia (1980-1992) can be undertaken over a 20 year period without apparent psychological or sociological harm. As a result, in their study, practically all women in Montreal who were referred for prenatal diagnosis of β -thalassaemia major or TSD had undergone screening in high school. The benefits include enabling informed reproductive choices to be made by those who, in a pre-screening era, may have been ignorant of their genetic carrier status. There has been a reduction in the frequency of these disorders in Montreal of 85%.⁶⁻⁸

After careful consideration of the strategies for optimising informed participation in community genetic screening programmes that have been trialled internationally,²⁻⁸⁻¹¹ and in consultation with Jewish community leaders, the ACGP decided that senior students attending high schools operated by the Jewish community were the optimal group for targeted screening in the TSDP.

While recognising the demonstrated benefits of a community genetics screening programme targeting adolescents such as that offered in Montreal, a number of authors and national and international guidelines produced by professional bodies and governments have urged caution in their implementation. Carefully evaluated pilot studies with attention to the psychosocial aspects have been called for.⁶⁻⁸⁻¹³ The study of the Sydney high school programme described in this paper is an example of such an evaluation.

Key points

- A genetics carrier screening programme for Tay-Sachs disease (1995-1998) and cystic fibrosis (1998) was developed in consultation with the local community targeting 817 senior high school students (15-17 years) attending four metropolitan Jewish community schools in Sydney, Australia.
- The programme comprised mandatory on site education followed by voluntary on site genetic carrier testing.
- A total of 629 students (77%) participated in the evaluation using anonymous coded longitudinal questionnaires to assess knowledge, attitude, and self-reported concern regarding a positive test result for carrier status, implemented pre- and immediate post-education and 12 months later. Measures also included uptake of testing and a survey of a sample of students who had tested three to six years later.
- The mandatory education session provided all senior students, regardless of whether they studied biology, with information on which to make a decision about testing, led to an increase in the number of students with a positive attitude towards genetic testing, and addressed concerns regarding carrier status. Confidentiality and autonomy were embedded in the programme design that resulted in a testing uptake of 94% by 1998.
- Three to six years later, there was a high retention of knowledge, low concern, high levels of satisfaction, and no stigma was experienced by genetic carriers, who reported positive intended result use.

This paper reports on the evaluation and impact of the programme with high school students who attended the education sessions conducted in participating high schools from 1995-1998.

METHODS

Setting and sample

In 1995, the Principals of the four Jewish schools in Sydney approved a mandatory education session during school hours. It was to be initially offered to all students in their final two years of high school (years 11-12, aged 15-18 years) at a time most suitable for the school timetable and followed by on site voluntary genetic testing for Tay-Sachs genetic carrier status two to four days later. After this first year, the programme was available only for students in their penultimate year of high school (year 11 students, aged 15-17 years).

In 1998, the programme was expanded from TSD to include cystic fibrosis (CF) also following similar consultations with the Jewish and school communities.

Table 1 Scales used to assess knowledge, attitude, and concerns

Knowledge of TSD (agree/unsure/disagree)	Knowledge of CF (agree/unsure/disagree)	Feelings if a genetic carrier (a lot/a bit/ not at all)	General attitude and use of test results (agree/unsure/disagree)
TSD affects the nervous system and the children die very young	CF affects the respiratory and digestive system	Worried about your own health	Support genetic carrier testing in high school
Babies with TSD healthy at birth	Treatment available for people with CF	Angry	Support genetic carrier testing for community
Ashkenazi Jews at higher risk of being genetic carriers of TSD	Ashkenazi Jews at higher risk of being genetic carriers of CF	Depressed	If both parents genetic carriers, testing in pregnancy should be offered
Genetic carrier will not develop symptoms of TSD	Genetic carrier will not develop symptoms of CF	Scared	Would encourage partner to be tested if found to be a genetic carrier
Both parents must be carriers of a TSD mutation to have a baby with TSD	Both parents must be carriers of a CF mutation to have a baby with CF	Less confident about forming relationships	
If only one partner is a carrier of a TSD mutation, there is no chance that will have baby with TSD	One parent can still be a CF genetic carrier even if the couple are told that they are not at risk for having baby with CF		
Faulty genes more common in certain populations			
Everyone a carrier of mutated genes			

Ethics approval

The study was subjected to review and approval by Institutional Ethics Committees of the investigators and by the Principals of all four high schools operated by the New South Wales Board of Jewish Education (Sydney, NSW).

The education sessions

The content of the education sessions was developed with expert and community consultations (manuscript submitted). All sessions in the four year period were presented by a genetic counsellor or other expert health professional.

The sessions covered detailed clinical description of the condition(s) for which testing was being offered, as well as brief descriptions of three autosomal recessive conditions with high genetic carrier frequency in the Australian population, TSD, CF, and thalassaemia. Inheritance patterns, mutation carrier frequencies, impact of genetic carrier testing, as well as the systems for result reporting were also covered. The voluntary nature of participation in the screening was emphasised. To avoid possible stigmatisation of genetic carriers, the message that everyone is a carrier of several mutated genes was promoted.

Implementation of voluntary on site testing and consent 1995-1998

Testing was offered at school two to four days after the education session. All students, regardless of their choice of testing participation, were required to attend an individual on site interview session conducted by a trained health professional. Protocols (available on request from KB-S) for the interviews were developed by a genetic counsellor.¹⁴

Participation was voluntary and the interviewer was non-directive. Students who were 16 years of age and over and who elected to participate completed a written consent form and had immediate access to testing. Written parental consent¹⁵⁻¹⁶ was required for any student not yet aged 16. Students who declined to participate were provided with written material describing how they could access testing at any future time.

Testing

The testing laboratory was accredited for medical testing,¹⁷ as required by Australian regulations and participated in the TSD International Quality Control Program.² Blood samples were used for all testing.

Testing for TSD was performed on venous serum or leucocytes, using initial *HEXA* enzyme analysis^{2-3,18,19} 15% of

appropriate specimens were also analysed by PCR to detect the three major DNA mutations of *HEXA*, resulting in a diagnostic sensitivity of between 92-98%.¹⁹ As this testing could also identify genetic carriers of Sandhoff disease, a disorder in which absence of both *HEXA* and *HEXB* is associated with a neurological phenotype identical to TSD,²⁰ students were informed of this possibility and provided with information at the time of consent. Sixteen *CFTR* mutations were analysed by PCR with a predicted diagnostic sensitivity of 95% for the Australian Jewish population.²¹

Result reporting

Both anonymous couple based (called scheme A)²² and individual (called scheme B) result report systems developed in the pilot programme¹ were offered to students in 1995 and 1996. From 1997, in response to identified barriers to testing uptake (see Discussion), a further system was developed and offered, called scheme D as it involved "Deferred" reporting of the test result (manuscript submitted). In scheme D, all subjects initially were registered anonymously and were issued a personal identification number ("PIN"). At any future time, a subject enrolled in scheme D could contact the testing programme and request to be transferred to either scheme A or B. Those transferring to scheme A were issued with a new PIN suitable for anonymous couple based reporting,²² while those transferring to scheme B were issued with a report of their results.

Students electing to receive their results and whose test result was negative were provided with a written report. However, if the student elected to receive their result and this was positive, initial telephone contact was made by a genetic counsellor or clinical geneticist associated with the programme, followed by a written report. However, all subjects were offered free access to a genetic counsellor, irrespective of which result reporting scheme they chose and of their test results. Students were encouraged to consult their doctor or seek rabbinical advice if they had concerns.

Testing was free of charge in 1995 if the students chose to have their results reported using scheme A, as only that particular system of result reporting was initially financially supported by the Jewish community. A charge (\$AU60) was levied if the student chose scheme B. However, once we determined that this practice was impacting on students' free choice (see later), this practice was discontinued and testing was provided free of charge to the student from 1996 onwards, regardless of their result reporting scheme choice.

Table 2 Percentage with scores of good knowledge of TSD (1995-1998) and CF (1998 only)

	T1	T2	T3 (test)	T3 (no test)
1995 (TSD)	21	95	79	69
1996 (TSD)	–	92	81	72
1997 (TSD)	–	96	85	73
1998 (TSD)	22	93	80	68
Significance of correlations				
Gender (p)	0.592	0.194	0.000	0.063
Studying biology (p)	0.013	0.350	0.014	0.054
1998 (CF)	31	94	89	72
Significance of correlations				
Gender (p)	0.084	0.063	0.000	0.081
Studying biology (p)	0.005	0.083	0.172	0.074

Values of $p < 0.05$ are shown in bold.

Evaluation measures

Anonymous coded longitudinal questionnaires¹⁴ were designed to assess knowledge, attitude, feelings, and reasons for testing choice. Students were informed of the purpose of the research and asked to complete the pre-questionnaire (T1) after they had entered the room for the education session but before the session began. To ensure that any change in knowledge, concern, or attitude was the result of the educational intervention alone, and to determine if good knowledge score was sufficient for making a decision regarding testing, the students were asked to complete the post-questionnaire within 10 minutes after the education session, before they left the room (T2). Twelve months later, the third questionnaire was usually completed in class (T3). The researchers knew only the code number for the questionnaires while the school held the link between the code and the students' identity, thus enabling students' responses to be serially tracked over the three time periods while blinding the investigators to the students' identity. This promoted the principle of confidentiality as students were asked to report their test result at T3. Socio-demographic data collected included age, gender, ancestry, and whether they were currently studying biology.

The scales used are shown in table 1. Knowledge of TSD from 1995-1998 was assessed from an eight item scale; the scale for CF used in 1998 comprised six questions. Attitude towards genetic testing in general and use of the results was assessed from a four item scale. A list of five adjectives or phrases were developed from findings in focus groups, conducted with students in 1995 to develop the education sessions (manuscript submitted), to describe their predicted (T1, T2) and reported (T3) concern if found to be a genetic carrier for TSD and or CF. The methodology for assessing levels of concern was based on the development of the Feelings About Test Results Measure that has been used previously in studies into the psychosocial impact of genetic testing for CF.²³ Students were asked to rank their feelings of concern from "a lot" to "not at all".

Reasons for testing choice were assessed from a seven item scale developed from the focus groups in addition to other reasons stated by the students during the evaluation period. Specific questions were also included at various times during the period of evaluation to determine the efficacy of education in addressing issues raised in focus groups and discussion with students, such as the understanding of the term "carrier".

In 2001, the questionnaires used at T3 were mailed to those students three to six years later who had agreed to participate in further research and who had by then already received their test result. These questionnaires were not anonymised in order to determine if the respondent knew what they had been tested for and if they correctly reported their result. Several questions were also included to examine their experiences and

views following participation in the genetic carrier testing programme.

Uptake of testing was measured by the proportion of students who had participated in the mandatory education and who subsequently elected to access the voluntary on site testing.

Analysis

Answers to the knowledge and general attitude questions were transformed into dichotomous variables. Correct knowledge answers were coded as 1, incorrect or unsure as 0. A maximum score of 8 was possible for knowledge of TSD and 6 for CF. Agreement with each general attitude question was also coded as 1 and classed as a "positive" attitude answer, with a maximum score of 4. For concern, "a lot" was transformed to a value of 6, mid-range to a value of 3, and "not at all" to zero: a maximum score of concern was 30.

For each scale, the baseline scores (T1) were a normal distribution enabling the further transformation of the scores to give a measure of good knowledge, positive attitude, and high levels of concern as greater than the median for the scale.²⁴ A good knowledge score was therefore assessed at 5 or more for TSD and 4 or more for CF and a positive general attitude was assumed if the score was 3 or more. High concern was assessed if the score was 16 or more.

Questionnaires transformed as above were analysed by Student paired *t* tests, Pearson's chi-square test, or Mann-Whitney tests as appropriate. Multiple linear regression modelling was used to explore associations and correlations among continuous variables. The significance level was set at 0.05, two sided.

Reliability of the scales was assessed by repeated application of the questionnaire one week later with the focus groups used to develop the education session. Internal consistency was assessed using Cronbach's Alpha. Validity of core questions on knowledge and attitude were determined by expert consultations.

RESULTS

Between 1995 and 1998, 817 year 11 students participated in on site education sessions before being offered on site testing, representing 93% of the eligible student population at the four schools, with 99.8% of respondents identifying themselves as Jewish. A total of 629 (77%) of these students participated in the evaluation. In 1995 and 1998, 283 students completed baseline questionnaires at T1 and questionnaires at T2 and T3 to determine the short and long term effects of the educational intervention and programme variations. In 1996 and 1997, 346 students completed questionnaires only at T2 and T3 to assess further decision making and the impact of programme variations. Overall, 51% of students participating in the evaluation were female, 27% were studying biology as a

Table 3 Percentage of students with a positive attitude towards genetic testing

	T1	T2	T3 (test)	T3 (no test)
Year				
1995	50	79	92	79
1998	73	84	95	76
Significance of correlations				
Gender (p)	0.032	0.196	0.081	0.009
Studying biology (p)	0.450	0.095	0.074	0.172

Values of $p < 0.05$ are shown in bold.

Table 4 Percentage of students with high level of concern about receiving a positive genetic carrier test result for TSD and CF (TSD/CF)

	T1	T2	T3 (test)	T3 (no test)
1995 (TSD)	52	20	5	23
1996 (TSD)	–	23	0	21
1997 (TSD)	–	21	3	24
1998 (TSD)	32	22	5	19
Significance of correlations				
Gender (p)	0.241	0.072	0.063	0.007
Studying biology (p)	0.092	0.350	0.421	0.063
1998 (CF)	34	17	3	14
Significance of correlations				
Gender (p)	0.352	0.092	0.081	0.009
Studying biology (p)	0.043	0.213	0.074	0.172

Values of $p < 0.05$ are shown in bold.

subject at the time of the education session; 15 were 15 years of age, 610 were aged 16 years, and four were aged 17 years at the time of testing.

Reliability of questionnaire

The component scales of the core questions covering knowledge and attitude were internally consistent ($\alpha = 0.77$ for knowledge and $\alpha = 0.55$ for general attitude).

Educational outcomes

Knowledge

There was no significant difference between baseline (T1) scores of good knowledge of TSD measured at 1995 and 1998 ($p = 0.247$), but more students had a good knowledge score for CF at T1 than for TSD ($p = 0.003$). There was no difference in good knowledge scores between the two conditions thereafter (table 2).

For both TSD and CF, there was a highly significant increase ($p = 0.000$) in good knowledge scores immediately following the education session at T2 (table 2). While studying biology was the only predictor of a good knowledge score at baseline, the only predictor at T2 was attending the education session (table 2). At this time, there was a highly significant correlation with a good knowledge score and choosing to use testing ($p = 0.000$ for TSD and $p = 0.002$ for CF). For both conditions, a good knowledge score was achieved by 99% of students who chose to test; 95% and 96% of the students who chose not to test had a good knowledge score for TSD and CF respectively.

Those who had testing had retained more knowledge 12 months later than those who had not had testing ($p = 0.012$ for TSD and $p = 0.008$ for CF) (table 2). For TSD, being female (85% F/72% M) and studying biology (79%/59%) were predictors of a good knowledge score at this time if they had chosen to test. However, for CF, being female (88% F/75% M) was the only predictor of a good knowledge score at this time if they had chosen to test. Correlations with gender or studying biology and a good knowledge score were not observed if the students had not had testing.

At T3, 73% reported that they recalled that they had had testing for TSD. Of the 44% who reported that they had

accessed their result, all stated their result, and 81% had good knowledge, compared to 72% who had not chosen to have testing ($p = 0.000$) (table 2). Of the students who identified themselves as genetic carriers of TSD, 10/11 (91%) had good knowledge, significantly higher than the 84% of students who were non-carriers ($p = 0.013$).

Of the student cohort in 1998, 27% reported transferring to scheme B and accessing their results for CF. Three stated that they were genetic carriers for CF. All had a good knowledge score compared to 84% of non-carriers.

General attitude towards genetic testing and its use

The percentage of students who had a positive attitude at T1 significantly increased between 1995 and 1998 ($p = 0.009$) (table 3). Immediately after the education session, a significant increase from baseline in the percentage of students with a positive attitude was observed ($p = 0.008$) (table 3). Being female (F 59%/M 41%) was a predictor of having a positive attitude at T1 but attending the education session was the only predictor at T2 (table 3); 96% of students who chose to test and 84% who did not have testing had a positive attitude ($p = 0.011$).

Students who had testing had a significantly increased positive attitude towards genetic testing over 12 months ($p = 0.024$) but there was no significant change if they did not have testing ($p = 0.061$) (table 3). Of those who did not test, females were more positive about genetic testing than males (74% F/34% M).

All 11 TSD genetic carriers reported at T3 that they would inform a future partner of the result, would use it in planning a pregnancy, and had told family members of their result. All three CF genetic carriers reported at T3 that they would inform a future partner of the result, two to three would use it in planning a pregnancy, one was unsure, and all reported that they had informed their family of the result.

Concern about having a positive test result

From 1995 to 1998, there was a significant reduction in baseline scores of high concern score regarding receiving a positive genetic carrier test result for TSD ($p = 0.012$) (table 4).

Table 5 Influences on testing choice (students could choose more than one reason). Results given as % of students

	1995	1996	1997	1998
Reason for testing				
To use with future partner	56	73	71	75
Just want to know	30	26	31	24
Testing now but results later	N/A	N/A	27	21
Advice of the Jewish community	46	7	5	4
Parents' influence	9	21	17	14
Peer pressure	6	9	3	1
Reason for not testing				
Not at this time of life	43	46	12	13
Hate needles	28	22	34	31
Don't believe I am personally at risk	20	22	24	23
Did not want to know carrier status	17	10	5	4
Cost	30	N/A	N/A	N/A
Test outside school	14	8	0	1
Parents' influence	10	11	10	10
Not in a high risk group	3	4	1	2

At T1 in 1998, there was no significant difference between having a high concern score for TSD compared to CF ($p=0.153$) (table 4).

Immediately after the education session there was a highly significant reduction in the percentage of students with a high concern score in both cohorts ($p=0.000$) (table 4). However, at T2 in 1998, the percentage of students with a high concern score for TSD was significantly greater than for CF ($p=0.001$), although this difference had disappeared by T3 regardless of their testing choice (table 4).

Studying biology was the only predictor of a high concern score at baseline for both conditions, but no correlation was found at T2 with a high concern score, including a decision to have testing ($p=0.075$ for TSD and $p=0.084$ for CF) (table 4). At T2, 23% and 47% of students who had poor knowledge had high concern scores regarding TSD and CF respectively, compared to 13% and 17% of students with a good knowledge score ($p=0.000$ for both TSD and CF).

Twelve months later, the percentage of students with a high concern score for both conditions had decreased if they had had testing ($p=0.000$) (table 4). For TSD, 2% of those who knew their result and 8% who had not accessed their result still had high concern scores; for CF no student who had tested and knew their result (carrier or non-carrier) had a high concern score compared to 4% who had not accessed their result. Where the students had not had testing, the high concern score for both conditions had not changed in 12 months ($p=0.061$ for TSD and $p=0.085$ for CF) (table 4). More males than females who had not had testing had high concern (TSD 32% F/67% M, CF 24%F/76%M).

Reasons for testing choices

Students were influenced by a number of factors (table 5) when making their testing choice. Charging for testing was clearly an influencing factor. In 1995, the Jewish community funded testing only if the student chose to enrol in scheme A and 46% of students reported that they chose this result reporting scheme (rather than scheme B) only because it was free. From 1996 onwards charges were no longer levied resulting in a marked rise in testing uptake (fig 1).

Students did not perceive that knowing their carrier genetic testing result with its reproductive implications was relevant to them at this time in their life, affecting testing use. Removal of this barrier in 1997 by the introduction of the "deferred" result reporting system called scheme D resulted in an increase in uptake (fig 1).

After removal of the above barriers, a fear of needles remained the major barrier to uptake. Males were more likely

Figure 1 Uptake of genetic testing 1995-1998. Schemes A, B, and D are the three reporting schemes described in the text. For 1997-1998, schemes A and B represent those students originally enrolled in scheme D who have elected to transfer to schemes A and B, while scheme D represents those students who have not yet made an election.

than females to state that they did not believe that they were personally at risk for being a genetic carrier even though they knew they were in a high risk group (66% compared to 24% respectively, $p=0.013$). There were no other significant differences in gender in their reasons for testing choice.

Testing uptake

Uptake for testing increased from 54% of eligible students in 1995 to 94% in 1998 (fig 1). Overall, more females had testing than males (64% compared to 46%). Recalled uptake for testing measured at T3 was not significantly different from the actual uptake ($p=0.461$). Moreover, the female/male ratio of recalled uptake was not significantly different from actual uptake ($p=0.101$). In 1998, all students who chose to have testing had testing for both TSD and CF.

In the four year period, only one parent (a lawyer) contacted the programme with a complaint. This parent objected to the fact that their child, rather than the parents, had been given a TSD genetic carrier test result. We explained that the student was legally able to give consent for the test¹⁵ and that such consent constituted a contract with the student and not the parent. The parent accepted this explanation.

In 1997-1888, all 371 students who had testing were initially enrolled in the "Deferred" result reporting system (scheme D). By July 2002, 163 (44%) had contacted the laboratory (now called the GeneTrustee, manuscript submitted) to transfer to schemes A or B, 65% of whom were female. The genetic carrier test results of 208 students therefore remained in scheme D anonymously linked to their PIN held by the GeneTrustee.

Long term follow up

Of the 273 students who had had testing and knew their individual result, 78% had agreed to participate in further research and were sent a follow up questionnaire in 2001. A total of 86 responses (44%) were received from those 196 students who could be contacted; 64% were from females. Eight students were genetic carriers for TSD (five females and three males) and two for CF, both females.

Three to six years after testing, 74% of the respondents had retained good knowledge of the conditions for which they had genetic carrier testing (table 6). There was no significant difference in gender ($p=0.082$); 90% of the genetic carriers had a

Table 6 Good knowledge scores about TSD and CF 3–6 years later

Year	% with good knowledge score
1995 (TSD) n=18	68
1996 (TSD) n=22	72
1997 (TSD) n=25	81
1998 (TSD/CF) n=21	75/86

good knowledge score compared to 63% of the non-carriers. No respondent, regardless of their test result, had a high concern score. No student incorrectly reported either their test result or what had been tested for. However, three non-carriers for TSD tested in 1995–1996 thought they would need to have testing again before planning a pregnancy and one respondent from 1996 questioned the need for a confirmation of her TSD genetic carrier test result before pregnancy. Ninety-one percent of the sample reported that they were either satisfied or very satisfied with the programme.

No stigma had been experienced by any of the genetic carriers. All had informed their extended family, 4/10 had told their family doctor, and 9/10 had informed close friends. Cascade testing was undertaken by other family members of 7/8 TSD genetic carriers (one was unsure) and of both CF genetic carriers. In the case where cascade testing had not been used, comment was made that testing would be taken up at school in future years when it was offered to her sibs. All of the genetic carriers agreed with the statement that “everyone is a carrier of several of mutated genes”; 93% of the non-carriers agreed with this statement, none disagreed, and the remainder were unsure, all of this latter group being from the 1996 testing cohort. In that year the emphasis on this issue was not as strong in the education session as in other years.

All genetic carriers indicated positive intended use and stated that they would inform their partner of their carrier status, encourage their partner to be tested, and, if their partner were also a genetic carrier, they would consider reproductive options and would seek genetic counselling.

DISCUSSION

Early screening is only effective if the information is used appropriately. As seen with the debacle of the sickle cell anaemia screening programme, and more recently with the inappropriate use of genetic test results in insurance, ignorance of the meaning of genetic test results can mean that carriers have a loss of self-esteem as well as social stigmatisation and discrimination.^{25–26} This study of the Sydney high school programme provides an opportunity to evaluate the psychosocial impact of implementation strategies for community genetic screening.

Concerns that are raised in the United States about possible discrimination in health insurance,²⁷ which may limit the design and evaluation of such a pilot programme, are not applicable in Australia (or in Canada). These countries have universal health insurance and thus allow investigation of issues of beneficence, autonomy, and informed consent without influence of insurance risk modifying free choice.

The Montreal study of Mitchell *et al*⁷ showed at a “macro” level the long term effectiveness and use of community genetics screening programmes within the high school population. Our present study, based on high school students in Sydney, Australia, extends their conclusions by allowing controlled evaluation of implementation strategies at a “micro” level. Our study design used anonymous, coded questionnaires, enabling longitudinal study of individual student responses within a cloak of confidentiality. This has enabled us to test the

effectiveness of specific interventions and to identify barriers to participation and to isolate key success factors.

The measure of success of the programme was not simply in the uptake of testing, although that was obviously desirable and important to the community. Its success was also that it provided awareness to the relevant community that Ashkenazi Jews were at increased risk for being genetic carriers of TSD and CF, an understanding of the conditions, and what it meant to be a genetic carrier for them, that testing was available at school or later in life, the importance of testing pre-pregnancy to enable reproductive choice, and the implications of being found to be a genetic carrier. In addition, understanding that everyone had several mutated genes was essential to minimise possible stigmatisation of identified genetic carriers. The education session and the programme design met these criteria and ensured that even those students who did not choose testing at school had raised awareness of testing availability for the future.

Uptake should ideally only occur after informed consent for testing, and this required adequate knowledge and uncoerced choice. As a result, evaluation of the effectiveness of educational interventions was necessary to ensure consent was both freely given and informed. We considered carefully whether there existed an implicit (and unintentional) element of coercion.²⁸ This may well be unavoidable in a group testing environment but every effort was made to limit the potential for coercion, with emphasis on the voluntary nature of participation in the education session, in the written material, and the use of a one on one consent interview to minimise the potential for peer pressure. The above notwithstanding, 5% of students indicated that peer pressure had influenced their testing choice.

We assessed baseline knowledge by questionnaire T1 in 1995. There was poor baseline knowledge about TSD in a community that might be expected to have good knowledge. The low baseline level remained when reassessed at T1 four years later in 1998, despite the annual implementation of the programme. It is therefore imperative that the educational programme is continuing and implemented annually. Baseline knowledge of CF, although still low and similar to that found in Canadian students,²⁹ was higher than for TSD, perhaps reflecting the fact that children with CF are now active in the community, and perhaps also reflecting the attention given to CF in the biology curriculum.

The differences observed between responses to the pre- and post-educational intervention questionnaires at T1 and T2 (administered only one to two hours later) allowed us to isolate and assess the impact and effectiveness of educational intervention. We assessed longer term educational impact by a survey 12 months later at T3 and three to six years later at T4.

Good knowledge and a positive attitude are important contributors to enabling informed choice about testing.²⁴ Ninety-nine percent of students who chose to test in our study had good knowledge, strongly suggesting that the strategy of on site education followed by one on one interview within the school curriculum succeeded in imparting sufficient knowledge to enable informed consent. It was also successful in engendering positive attitudes towards genetic testing, and in lowering and allaying individual subjects’ concerns towards genetic testing and its potential impact on the subjects’ health.²⁹ Testing choice was not correlated with high concern scores.²⁴ Interestingly, although those students already studying biology had a higher level of knowledge (as might be expected), these students also had higher levels of concern towards genetic testing, suggesting that there may be a gap between objective knowledge and subjective interpretation of the significance of this knowledge, a gap that can be closed through direct participation in a genetic screening programme.

Those students who had participated in the genetic testing programme and elected to be tested showed better retention

of knowledge, and lower levels of concern, than those who declined to be tested. Students also appeared to understand the potential use of this knowledge and indicated their intentions to use this knowledge rationally in future reproductive decisions, a finding in keeping with Mitchell *et al.*⁷ In 1998, only 6% of eligible students did not take up testing. Of those students who reported at T3 that this was their testing choice, 95% had had a good knowledge score at T2, the time they were making the choice about testing. Therefore their negative choice was based on information.

Long term follow up of students three to six years after participation showed that knowledge was retained even for extended periods. Our findings are at some variance with reports of others, who had suggested rapid evaporation of knowledge.³⁰ The results obtained by us may be related to the design of the screening programme, with its intensive educational session by experts in the field, the encouragement by the schools and the community for wide spread discussion by students, family, and community,³¹ and the reinforcement of awareness of genetic testing through annual scheduling of the genetic education and testing as part of the school curriculum. However, in keeping with these same earlier studies,³⁰ we observed some decay in quality of retained information, with a few students at T4 misunderstanding the significance of a positive or negative genetic carrier result. As these particular students arose only from the earliest cohorts of the screening programme in 1995-1996, it is possible that continuing evolution and refinement of our educational programme may have addressed this in later years. In keeping with the findings of others,²⁹ we did not completely succeed in ensuring an equal gender appreciation of the significance of genetic testing, and more female than male students took up testing and subsequently accessed their results.

We also have shown that increasing the amount of information imparted, from TSD alone, to both TSD and CF, did not impact adversely on the students' understanding and ability to make informed decisions. However, given the fact that the CF test could be considered to have been "added on" to the TSD test, we have not yet shown that the students made an independent choice about TSD and CF and this will need to be addressed in further studies. It should also be noted that subjects in our present study were all from a genetically homogeneous pool with regards to the frequency distribution of different CF mutations.²¹ This meant that we did not have to consider the complex issue of different diagnostic sensitivities for CF carrier detection dependent on ancestry and ethnic origin.

Evaluation of factors nominated by students as blocking or discouraging their participation in the programme allowed us to identify three significant factors: cost, time of life, and fear of needles.

Cost was nominated as a strong influencing factor both for participation in the programme, as well as for election of the scheme by which results were to be reported. In order to ensure that there was no implicit coercion in free choice, it was necessary to offer testing free of charge; a corollary is that a fee for service strategy would have resulted in lack of equity and/or lack of free choice.

We identified a previously unsuspected, but not surprising, barrier to testing in that students felt that testing at 16 years of age was too early for them to associate this act with future reproductive planning. It has long been recognised that educators need to make their teaching relevant to the students and that they must also engage them to enhance their learning if this is to be used as the basis of decision making.³² When a strategy of "test now, but get results later in life" was offered through the development of scheme D deferred result reporting, participation and uptake rose to unprecedented levels of 94%, much higher than those reported by other major analogous voluntary secular community screening programmes.² While the development of scheme D is a promising new strat-

egy, it is as yet too early to know if the higher level of uptake will translate to an equally high level of test result use. As at the date of writing of this paper (2002), while 44% of scheme D results have been accessed, the student cohort has probably not yet reached the age at which all participants may have needed to use this information. A longer term follow up study is planned.

Finally, dislike of needles and venepuncture (table 5) was identified as a significant barrier to participation and this was also reported by Durfy *et al.*²⁹ Although current testing technologies may favour TSD laboratory testing requiring blood samples and thus necessitating venepuncture, newer laboratory techniques may allow migration of sample collection to mouthwash,^{33, 34} removing a major barrier to testing participation.

It is important to note, however, where the programme may be yet further improved. Three non-carriers thought they needed to have testing again before pregnancy. One student questioned the need for a confirmation of her TSD carrier test result before pregnancy. This case may be a form of denial, although she indicated that she had no concerns in any of the areas surveyed. On the other hand it may be a basic misunderstanding of the fact that a DNA test result does not change over time.

In addition, the single largest block of Australian Jews are unaffiliated and cannot be reached through Jewish schools alone.³⁵ Many Sydney students of Jewish ancestry do not attend the community Jewish schools and so the programme will need to be extended to ensure that the principles of access and equity are met. Testing the programme with other Australian Jewish communities will also be important.

Research is needed to determine if a programme that has been designed for the Jewish community may well be extended to other communities in multicultural Australia, as has been illustrated with thalassaemia screening in Montreal⁷ and Hong Kong.³⁶ The results of these studies will determine if the Sydney programme initiated with the Jewish community is a model community genetics programme for high school students.

CONCLUSION

Principles that guide a community friendly, sensitive, scientifically rational, and ultimately beneficial screening programme for the Jewish community have been proposed.³⁵ These state that addressing the concerns of the Orthodox community is essential, that consultation with community leaders, rabbis, activists, and prominent people is critical in programme planning, and the use of stereotyping of Jews only as those at high risk must be avoided. All of these, as well as the principles of autonomy and confidentiality and facilitation of access to appropriate professional and community information and support services, were embraced in the development, evolution, and implementation of the Sydney programme.

As stated in the 1995 film *Mr Holland's Opus*, "A teacher has two jobs. [To . . .] fill young minds with knowledge, but more importantly, give those minds a compass so that [the] knowledge doesn't go to waste".³⁷ The education session underpinned this community genetics programme, facilitated informed choice regarding testing and discussion of the implications of its use, and enabled retention of that knowledge for future use.

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