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ORIGINAL RESEARCH

Huntington Disease: Who Seeks Presymptomatic Genetic Testing, Why and What are the Outcomes?

Tracey M. Scuffham · John C. MacMillan

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Abstract The aims of this study were to: 1) quantify the characteristics of those seeking presymptomatic testing for HD, 2) identify their motivations for testing, 3) quantify the waiting times between the various steps within the testing process, and 4) quantify the outcomes of testing at a large state-wide genetic testing center in Australia. A review of medical charts for all referrals for presymptomatic testing of Huntington disease received over a 4 year period (2006–2010) was undertaken. A total of 152 cases met the study inclusion criteria; the mean age was 39 years, 46 % were male and 61 % underwent genetic testing. Of the males who were tested there was a non-significant trend towards having an affected mother vs father (62 %, $p = 0.09$), whereas females tested were just as likely to have an affected mother or father. The most frequently cited reasons for seeking testing were "family planning", "plan future", and "need to know". Some 11 % deferred testing following the psychological assessment. Of those at 50 % prior risk, 57.5 % tested positive; this was higher than expected and much higher than reported in other studies. The median times from referral to initial appointment, and then to results was 69 days and 144 days respectively. Overall, this review of medical charts shows the depth of information obtainable from routinely collected data and revealed that a high proportion of patients tested positive for HD at this centre.

Keywords Health services · Genetic testing · Outcome and process assessment (Health Care) · Alleles · Penetrance · Australia · Epidemiology · Predictive testing · Huntington disease

T. M. Scuffham (✉) · J. C. MacMillan
Genetic Health Queensland, Royal Brisbane and Women's Hospital, Brisbane, Queensland, Australia
e-mail: tscuffham@internode.on.net

J. C. MacMillan
Department of Medicine, University of Queensland, Brisbane, Australia

Introduction

Since 1994, presymptomatic direct genetic testing for Huntington disease (HD) has been available in Queensland, Australia. Queensland is the second largest and third most populous state in Australia. Genetic Health Queensland (GHQ) is a state-wide specialty service that provides publicly funded genetic services for a population of approximately 4.6 million (Australian Bureau of Statistics 2011). Genetic testing in public hospitals is financed through the State and Australian governments with funds raised by taxation (i.e. a form of social insurance for the population), free inpatient health care is provided to all citizens and there is minimal cost to patients for outpatient services and pharmaceuticals. Each year approximately 50 people are referred by a medical practitioner to GHQ for genetic counseling regarding presymptomatic, diagnostic, or prenatal testing for HD. Presymptomatic testing at GHQ is guided by guidelines from the Human Genetics Society of Australasia (HGSA) and international guidelines (Human Genetics Society of Australasia 2005; International Huntington Association and World Federation of Neurology Research Group on Huntington's Chorea 1994). Potential adverse consequences of genetic testing for HD include psychological distress from the knowledge the patient will develop the disease, changes in family dynamics, workplace and social discrimination such as an inability to obtain insurances in some countries, including the potential to be denied life insurance in Australia or incur increased costs for coverage. In 2013, the federal government introduced plans for publically-funded long-term disability insurance.

Whilst each individual is assessed at GHQ on a case-by-case basis, the typical process for presymptomatic testing involves at least four consultations with a multidisciplinary team. GHQ allocates up to 1 h for each consultation, however the actual duration of a session may vary. Additional consultations for genetic counseling are available if indicated and/or requested by the patient. A follow-up consultation with the psychiatrist/psychologist is recommended for all patients following testing.

Previous studies that have reported on the genetic testing process for HD include studies from Victoria (Australia), Quebec, and South Africa (Dufrasne et al. 2011; Futter et al. 2009; Sizer et al. 2012; Tassicker et al. 2006; Trembath et al. 2006). Those studies show there are some similarities and differences in the demographics of those who seek testing. Three recent comparative studies, from Johannesburg, South Africa, Quebec, Canada and the most recent Australian study from Victoria are shown in Table 1 as an example. These studies found that individuals seeking predictive genetic testing for HD tended to be in their 30s, female and approximately a third or more were childless. Two of those studies reported motivations for testing.

The time to proceed through the genetic testing process is rarely reported. One study of 756 participants reported a mean time from the time the patient became aware of their risk to the time of receiving their test result (9.7 years with a mean age of entering testing of 40 years) (Trembath et al. 2006). A prolonged process time may increase anxiety and distress whilst a rapid process time may not allow the patient sufficient time to fully consider the implications of testing and their possible reactions (Solden et al. 2000).

The outcomes reported from presymptomatic testing of those at 50 % prior risk are generally similar across studies with between 37.5 % and 40.0 % testing positive for HD (Dufrasne et al. 2011; Sizer et al. 2012; Trembath et al. 2006). A previous Australian study reported 38.1 % of those tested in Australia had 40 or more CAG repeats, of which 39.5 % from Queensland had 40 or more CAG repeats (Tassicker et al. 2006).

The Australian studies mentioned previously were based on data up to 2003 (Tassicker et al. 2006) and 2004 (Trembath et al. 2006), and included periods from before direct DNA analysis was available; therefore, it was of interest to assess recent patterns of HD presymptomatic testing for the state-wide service in Queensland and how these patterns compare with historic and international findings. Accordingly, this study sought to identify *who* gets testing, *why* they seek testing, *how* long testing takes, and *what* the outcomes were. The aims of this study were to: 1) quantify the characteristics of those seeking presymptomatic testing for HD at GHQ, 2) identify the motivations for testing, 3) quantify the waiting times between the various steps within the testing process, and 4) quantify the outcomes of testing at a large state-wide genetic testing center in Australia. The characteristics that differentiate those who approach a genetic service for testing from those who do not seek testing have been reported previously (Pakenham et al. 2004).

Methods

A review of medical charts in a large state-wide genetics service (GHQ) based in Brisbane at the Royal Brisbane and Women's Hospital (RBWH) was undertaken, and was approved by the RBWH Institutional Review Board (approval number HREC/10/QRBW/520). Cases were identified from a database of patients referred to GHQ for HD risk assessment and testing from February 2006 to 2010. The database was searched in February 2011; this allowed sufficient time between referral and the chart review for the majority of those interested in testing to have completed the testing process.

The medical chart contents reviewed by the genetic counselor included the referral letter, consultation dates, medical notes, pedigree, test results, and assessments and follow-up letters from the psychiatrist. Data extracted included the above information plus the source of referral, patient demographics, and motivations for testing. An Excel spreadsheet was developed for data entry of chart information.

Motivations were identified by reading the notes of the geneticist and genetic counselor for all consultations and up to three motivations were extracted for each patient. An initial random sample of 30 cases was used to identify patient's key

Table 1 Descriptive information of three relevant comparative studies

Country of study:	Johannesburg, South Africa	Quebec, Canada	Victoria, Australia
Authors (year)	Sizer et al. (2012)	Dufrasne et al. (2011)	Trembath et al. (2006)
Mean Age (years)	30.0	36.4	40.4
Gender (% female)	66.7	57.0	57.6
Childless (%)	54.4	43.0	31.5
50 % prior risk (%)	NR	98.5	88.8
40+ CAG repeats (%)	29.9	40.0	37.5
36–39 CAG repeats (%)	NR	3.7	3.2
Motivations for testing:			
Decrease uncertainty	42.5	34.1	NR
Family planning	5.5	16.1	NR
Plan future	34.2	15.1	NR
Inform children	12.3	14.8	NR

NR not reported

motivations; the full text was recorded and later summarized into the emerging themes for subsequent data extraction and analysis. The limit of three motivations was chosen as less than 1 % of the sample had more than three motivations recorded.

Patients who were tested for HD were compared with those who withdrew from testing. The number of CAG repeats are reported for those who had genetic testing. Those who had not returned for their results after testing were counted as tested.

Data Analysis

Data were coded and descriptive analysis undertaken. Waiting times between steps in the process were calculated to identify the lag times of the process and the overall time to obtain a result. Tests for differences between those tested and not tested were undertaken using Chi-square tests for categorical variables, and t-tests for continuous variables. Chi-square tests for differences in proportions and exact binomial tests for nominal variables with only two values (e.g. the proportion of males tested that have an affected mother with a null 0.5 probability of having an affected mother) were used.

Results

A total of 262 patients were identified from the database who were seen for HD. Of these, 110 cases were excluded due to: the initial referral was pre-2006 (n= 46), the patient was not seen (appointment cancelled by patient n= 13, or patient failed to attend n= 20), the patient was symptomatic and referred for confirmatory diagnostic test (n= 18), the "patient" was not at risk for HD but was seeking information because of their at-risk partner (n= 3), or there was no information in the chart (n= 10). Of the 33 patients who cancelled or failed to attend their appointment, 70 % were female and 30 % male (p= 0.02). Data were extracted for the remaining 152 cases, all of which were referred for presymptomatic genetic testing. The vast majority of referrals were from General Practitioners (GP's), followed by neurologists and other clinicians.

Of the 152 cases analyzed, 148 (94 of those tested and 54 of those not tested) had sufficient chart information to describe the timing between steps in the genetic counseling (and testing) process. Some 32 % of the cohort withdrew from the testing process following the initial consultation; primary reasons, for example, were that some sought information only (n= 12; 8 %) whereas others decided against testing or to defer genetic testing (n= 36; 24 %). Of those who had a consultation with a psychiatrist (n= 79), 11 % did not proceed further with the testing process (i.e. 6.6 % of the total cohort). This was usually a joint decision between the psychiatrist and patient. Some 15.0 % who provided a blood sample did not have a record of a psychological assessment.

Characteristics of Those who Seek Testing

The mean age at time of referral was 39 years, with a range from 4 years (where a parent was seeking predictive testing of their child) to 77 years (Table 2). Fewer males than females were seen (46 % vs 54 %) and the majority of patients were married (70.2 %; Table 2). Of those who were seen, 62 % and 60 % of the males and females respectively were tested. The majority of patient charts (72 %) had data on affected family members; 59 % and 52 % of males and females respectively had an affected mother. Of the males who were tested, 62 % had an affected mother (p= 0.094), whereas for females tested, the affected parent was just as likely to be the mother or father (49 % vs 51 %). For males and females not tested, the affected parent was just as likely to be the mother (55 %) or father (45 %). Those who underwent testing had more first-degree (1.41 vs 0.93; p= 0.005) and second-degree (1.39 vs 1.27; p= 0.045) affected relatives compared with those who were not tested. Overall, those tested had significantly higher prior risk than those not tested (p= 0.001). That is, 91 % of those tested had a prior risk of 50 % versus 68 % of those not tested; whereas 32 % of those not tested had a prior risk of 25 % or less compared with 9 % of those tested.

Motivations for Seeking Genetic Testing

A total of 150 motivations for genetic testing were identified from 123 patient charts which contained a motivation for testing. These were categorized into eight themes where the motivation was reported by at least 2 % of patients (Table 3). The most commonly occurring theme was "family planning" for both those tested and not tested, followed by "plan future", "need to know" and "inform children". Significant differences between those tested and not tested were identified for the proportions reporting "plan future" (p= 0.049) and to "obtain information" (p <0.001), with those not tested being more likely to have undertaken counseling to acquire information about the disease, their risk and/or testing process.

Timing of the Process

From the time of referral, 50 % of patients were seen for an initial consultation within 10 weeks, and 75 % within 4 months (Table 4). A psychological assessment was required after the initial consultation. Although the date of the psychological assessment was not recorded, the median time between the initial genetics consultation and the second genetics consultation, when a blood sample is taken, was 12 weeks (range: 0–115 weeks). The distribution around this factor was skewed by several large values and 75 % of patients were seen for their second genetics appointment within 21 weeks. Delays in this step of the process were largely patient driven; for example,

Table 2 Descriptive characteristics of the study sample

Characteristic	Total	Tested	Not tested	*p*-value[a] (tested vs not tested)
Total sample (N)	152	95	57	
Age (years) [mean (SD)] *(n= 152)*	39.3 (14.3)	41.0 (14.1)	36.5 (14.2)	0.464[b]
Gender [% male] *(n= 152)*	45.5 %	46.2 %	44.1 %	0.794
Location of residence (n= 151)				
Metropolitan Brisbane	32.9 %	34.8 %	31.0 %	
Regional Queensland	67.1 %	65.2 %	69.0 %	0.847
Gender of affected parent (n= 110)[c]				
Males (*n*= 54):				
Mother	59.3%[d]	62.2%[d]	52.9 %	
Father	40.7%[d]	37.8%[d]	47.1 %	0.522
Females (*n*= 56):				
Mother	51.8%[d]	48.6%[d]	57.9 %	
Father	48.2%[d]	51.4%[d]	42.1 %	0.512
Marital status (n= 141)				
Single	22.0 %	23.1 %	20.0 %	
Married	70.2 %	68.1 %	74.0 %	
Separated/divorced/widowed	7.8 %	8.8 %	6.0 %	0.734
Number of children (n= 147)				
% with no children	41.5 %	38.9 %	45.6 %	0.718
Mean (SD) children for those with 1 or more children	2.49 (1.3)	2.38 (1.10)	2.68 (1.68)	0.327[b]
Referral source (n= 152)				
GP	86.8 %	89.3 %	83.0 %	
Neurologist	4.6 %	3.2 %	6.8 %	
Other specialist	8.6 %	6.5 %	10.2 %	0.415
Number of relatives affected				
Mean (SD) 1st degree relatives affected *(n= 145)*	1.23 (0.9)	1.41 (0.9)	0.93 (0.8)	0.005[b]
Mean (SD) 2nd degree relatives affected *(n= 140)*	1.34 (1.4)	1.39 (1.5)	1.27 (1.1)	0.045[b]
Prior risk (n= 147)				
Prior risk of 50 %	82.3 %	91.2 %	67.9 %	
Prior risk of 25%[e]	4.1 %	2.2 %	7.1 %	
Less than 25 % prior risk[e]	13.6 %	6.6 %	25.0 %	0.001

[a] Chi-square test used unless otherwise indicated by superscript "b"

[b] Student's *t*-test used for differences between means

[c] For the 110, females and males with an affected mother and father were 26.4 %, 24.5 %, 29.1 % and 20.0 % respectively

[d] Of the male (*n*= 54) and female (*n*= 56) samples, the probability of finding 59.3 % males had an affected mother or 51.8 % of females had an affected mother were *p*= 0.110 and *p*= 0.448 respectively. Of those tested (37/54 males and 37/56 females), the corresponding probabilities having an affected mother were males: 62.2 %, *p*= 0.094 and females: 48.6 %, *p*= 0.62

[e] Prior risk of 25 % or less: Total=17.7 %, Tested=8.8 %, and Not tested=32.1 %

the reason cited in the chart of one patient was that they were waiting for a family member to also go through the counseling process and then for both of them to receive their results at the same time. Once the blood sample was taken, the time to the third appointment to receive the results was approximately 7 weeks (patients are contacted to make this appointment only when the results are returned from the testing laboratory, and results are given to patients at the next available appointment). Overall, the median time for the genetic testing process, from referral to result, was 32 weeks (Fig. 1), and from the initial appointment to result was 21 weeks.

Not all patients received their result—approximately 7 % (*n*= 7) of those tested had not returned for a final genetics consultation to receive their result during the study period. Four of these patients had been referred in 2006, one in 2007 and two in 2009. The mean age was 46.4 years (range 30–71), four were male, and four married. There were records of a psychological consultation for two of these patients only; and

Table 3 Motivations for seeking genetic testing

	Total n= 150/N= 123	Tested n= 104/N= 82	Not tested n= 46/N= 41	p-value[b]
Family planning	24.0 %	26.0 %	19.6 %	0.398
Plan future	20.7 %	25.0 %	10.9 %	0.049
Need to know	16.7 %	15.4 %	19.6 %	0.526
Inform children	16.0 %	18.3 %	10.9 %	0.254
Obtain information	8.0 %	2.9 %	19.6 %	<0.001
Diagnosis	7.3 %	5.8 %	10.9 %	0.269
Career	4.7 %	4.8 %	4.3 %	0.902
Other[a]	2.7 %	1.9 %	4.3 %	0.395

n = total number of motivations identified from those with at least one motivation identified

N = total number of case records with at least one motivation identified

[a] Other = "own health" concerns (n= 3) and to "help relatives" (n= 1)

[b] Chi-square tests for differences in proportions used

five lived in regional areas of Queensland. All were at 50 % prior risk, and five of these seven (71 %) reported having an affected mother. Five had CAG expansions in the disease-causing range and two in the normal range.

Outcomes of Genetic Counseling and Testing

The distribution of CAG repeats for those who underwent a genetic test are illustrated in Fig. 2 and categorized in Table 5. Overall, 54.6 % of those tested had CAG repeats in the disease causing range, including the 3.1 % with reduced penetrance alleles. Those with a 50 % prior risk were more likely to have CAG repeats in the disease-causing range (57.6 %) whereas 30 % of those with a 25 % or less prior risk had disease-causing CAG repeats. In addition, four patients had CAG expansions on both alleles; expansions on their shortest allele were in the intermediate range (27/43; 27/45; 28/42; 29/44). CAG repeats in the reduced penetrance range were more common in those with a prior risk of 25 % or less compared to those with a 50 % prior risk (p= 0.02). However, this was based on just eight patients.

For those with a 50 % prior risk aged less than 40 years, 63.8 % had CAG repeats in the disease-causing range (71.4 % in the 30–39 age-group had disease-causing CAG repeats) whereas for those aged 50 years and over, disease-causing CAG repeats were identified in 47.8 % (33.3 % in those 60 years and over). These differences were statistically significant (p= 0.005).

Table 4 Waiting times between stages in the process of genetic testing at GHQ

	N	Median (days)	Percentiles	
			25th	75th
Referral to initial appointment	148	69	33	114
Initial appointment to blood test	91	86	25	148
Blood test to results	84	49	35	63
Total days from referral to result	85	225	164	323
Total days from initial appointment to results	84	144	85	229

Discussion

This study presents a summary of experience with providing genetic counseling for individuals at risk for Huntington disease between 2006 and 2010 in Queensland, Australia. This study describes the characteristics, motivations, timing and outcomes of those referred to GHQ for presymptomatic testing. The key findings of interest from this study are the differences in motivations for testing and the high proportion of those who tested positive. These factors are discussed below.

Characteristics of Those who Seek Testing

It is possible that growing up with an affected mother may have a greater impact on the motivation to seek genetic testing than growing up with an affected father. In the sub-sample of N= 110 with information on the affected parent, 26.4 % and 24.5 % were females with an affected mother and father respectively, and 29.1 % and 20.0 % were males with an affected mother and father. Although these differences failed to reach statistical significance (p= 0.011), one more male with an affected mother instead of an affected father would have produced a statistically significant result, and therefore this finding warrants further consideration. Affected mothers may positively influence their sons to seek genetic testing given their typical care-giving role and/or fathers may deter sons from testing; however, these patterns were not seen in daughters. In this sample it was apparent that both effects occurred - males with an affected mother were more likely to seek genetic testing and males with an affected father were

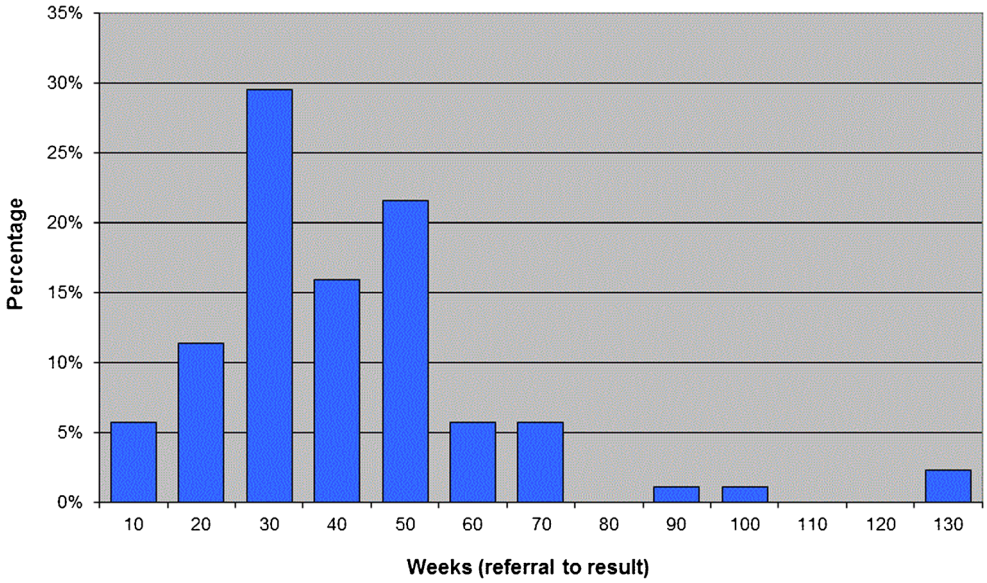

Fig. 1 Time from initial referral to receiving test result

much less likely. The psychological mechanisms behind this are unknown, however sons of affected fathers may simply not want to know, may feel more resilient to future risks (i.e. expect that developing HD won't happen to them), or may feel more resigned to following in their father's footsteps (i.e. expect to develop HD). No other study was identified that had reported on the gender of the affected parent and therefore it is not possible to determine whether this finding is typical or unique to this sample. If this is a common finding, further understanding of this phenomenon should be sought. Moreover, this finding may have implications for targeting sons of affected fathers for genetic testing given that males are less likely to seek genetic testing than females (see Table 1).

Motivations for Seeking Genetic Testing

The most frequently cited motivations for seeking testing in this study, in rank order, were family planning, planning for the future, a "need to know" (i.e. "reduce uncertainty") and to inform children. Family planning was the third and fourth most cited reason in the Quebec (Dufrasne et al. 2011) and Johannesburg (Sizer et al. 2012) studies respectively. Both the Quebec and Johannesburg studies cited a "need to know" (reduce uncertainty) as the most common reason for seeking testing but was ranked third in this Queensland sample (recorded in 17 % of cases). Those wishing to "get rid of uncertainty" as their reason for testing are likely to be more distressed, both pre- and post-testing, and this reason for testing may be an indicator of long-term distress (Decruyenaere et al. 2003). Thus, knowing the patient's motivation provides an opportunity to assist the patient with determining if, and when, they are ready to proceed with genetic testing. It also enables the genetics team to provide support as well as specific and relevant information to ensure patients are informed in their decision-making.

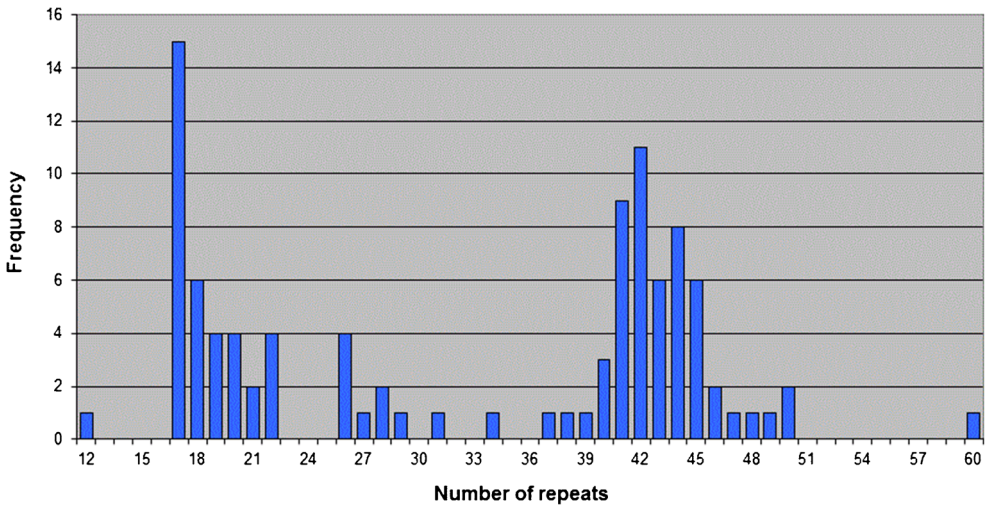

Fig. 2 Distribution of CAG repeats

Table 5 Allele sizes

CAG repeats on the longest allele		50 % prior risk (n= 85)	Prior risk ≤ 25 % (n= 10)	Total (n= 95)
≤ 26	Normal range	36.5 %	60.0 %	39.2 %
27–35	Intermediate range	5.9 %	10.0 %	6.2 %
36–39	Reduced penetrance	1.2 %	20.0 %	3.1 %
≥40	Disease range	56.5 %	10.0 %	51.5 %

Timing of the Process

A key feature of the present study was to document and quantify the timing between various stages in the genetic testing process. No other study was identified that reported timing of the various stages within the testing process. Of note was that the 3-month lag between the initial appointment and the appointment when the blood sample is taken was the longest interval in the testing process. This lag enables the patient time to fully comprehend the implications of the test and to be sure they wish to proceed. Moreover, the patient is required to undergo a psychological assessment in this period. Interestingly, 32 % of the cohort withdrew following the initial consultation and an additional 6.6 % of the total cohort withdrew after undergoing a psychological assessment. The withdrawal rates in this study were much greater than those of an earlier comparable Australian study where 8 % withdrew after their first consultation and another 6.3 % withdrew later in the testing process (14.3 % overall withdrew) (Trembath et al. 2006). However, in that study psychological assessment (or neuropsychiatric assessment) was not required and was declined by 28 % of that sample (Trembath et al. 2006). A recent Canadian study reported that 11.6 % overall withdrew from testing after psychological counseling (Dufrasne et al. 2011); this is greater than the present study for that stage in the testing process and may be due to differences in attitudes towards testing, and/or healthcare systems. These withdrawal rates are substantially different between studies, especially when the point of withdrawal is taken into account.

Outcomes of Genetic Counseling and Testing

In this study, 56.5 % of those with a 50 % prior risk (71 % in the 30–39 age group, 33 % in the ≥60 age-group) tested positive (CAG repeats ≥40) which is a higher percentage compared to the 30–47.6 % reported in previous studies (Dufrasne et al. 2011; Futter et al. 2009; Sizer et al. 2012; Tassicker et al. 2006; Trembath et al. 2006). Similarly, for those with a 25 % prior risk, 30 % in this study tested positive compared with the 8–27 % in other studies (Creighton et al. 2003; Harper et al. 2000; Tassicker et al. 2006). The relatively high rate of positive test results in this study may be due to several differences between this study and other studies, such as the age at presentation, motivation for seeking testing, the recent data used in this study (see Tables 1 and 2) and chance.

An interesting finding was there were four patients who had CAG expansions on both alleles (approximately 5 % of those tested). These four patients all had CAG repeats in the disease causing range on the longest allele and CAG repeats in the intermediate range on the shortest allele. CAG repeats in the intermediate range are meiotically unstable and may expand to cause HD in subsequent generations (Chong et al. 1997; Hogarth 2003; Semaka et al. 2010). A recent intergenerational study of patients with intermediate CAG repeats reported that 14 % had expanded into the disease-causing range, and 9 % contracted, on transmission to offspring (Semaka et al. 2010). Thus, any offspring from one of these patients may have a higher risk of developing HD than the general population even if they inherit the shortest allele.

Limitations

There are a number of limitations with this study. The relatively small sample of 152 cases limits the generalizability of results; however, all available data was obtained for all cases referred to a large State-wide genetics service in Australia over a recent 4-year period. There were some missing data, and some cases had minimal or no usable data recorded in the patient charts. Of note was the recording of patient motivations for testing; some patients had up to four motivations identifiable from their charts and some (19 %, 29/152) had no motivations recorded.

There is potential to have assigned some cases to the "not tested" category as a census of 12-months post referral was used. Thus, some participants seen in 2010 or earlier may not have had sufficient time to make a decision about continuing with the testing process. Some patients may come back years after an initial appointment to proceed with genetic testing. Similarly, of those tested, seven (7.7 % of those tested) did not return to obtain their result.

One limitation in the analysis of timing around the service was the non-recording of the date the patient was seen for their psychological assessment. This was partly an oversight during the data extraction process. However, psychiatry/psychology is a different department within Queensland Health and GHQ has no direct control over their waiting lists. As such, that waiting period for the patient journey is not modifiable by GHQ. There was generally no information contained in patient charts for any follow-up psychological consultations, and therefore, the proportion of patients who sought further psychological support for adjustment to results is unknown.

Conclusion

This study described the patient characteristics and outcomes of patients referred to a large State-wide genetics service for presymptomatic HD testing over a 4-year period. The proportion who tested positive for HD was substantially higher than previous studies. The waiting times between steps in the process provides a baseline indicator of the responsiveness of genetic services allowing further studies for monitoring and international comparisons.

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Conflict of interest statement The authors, Tracey Scuffham and John MacMillan, declare that they have no conflict of interest.

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