## ABSTRACT

Huntington Disease (HD) is a neurodegenerative disorder that is inherited in an autosomal dominant manner, and for which testing is available. The aim of this retrospective file-based study was to analyse the numbers and demographics of individuals who had diagnostic, predictive or prenatal genetic counselling and/or testing for HD between January 1998 and December 2006 through the Division of Human Genetics, National Health Laboratory Service and University of the Witwatersrand, Johannesburg.

Files for 287 individuals who had genetic counselling and/or testing for HD were included in this study, with 77% being diagnostic cases, 20% predictive and 3% prenatal. When the results obtained in this study were compared to a study by Kromberg et al. (1999) done previously in the same Division, it was found that there has been an increase in the number of diagnostic and predictive tests done per year during this study, with diagnostic tests making up a greater percentage of the total number of tests performed.

One of the objectives of this study was to characterise the individuals who requested HD testing and to compare the characteristics of those in the diagnostic testing group to those in the predictive testing group. The median age of the individuals in the predictive testing group was 30 years, which was significantly different from the median age of 49 years for individuals in the diagnostic testing group ($p<0.001$). It was found that there were significantly more women than men requesting predictive testing ($p=0.02$), while the number of males and females in the diagnostic testing group was similar ($p=1.00$). There was also a greater percentage of employed (76.4%) versus unemployed (23.6%) individuals in the predictive testing group, while the percentages of employed and unemployed individuals in the diagnostic testing group were similar (45.5% and 54.5% respectively). Significantly more individuals in the diagnostic testing group had children (74.5%) compared to those in the predictive testing group, where 44.6% of individuals had one or more children. There was a greater percentage of white individuals in the predictive testing group (91% white; 3.5% black) compared to the diagnostic testing group (48% white, 42% black).

The completion rate of the predictive testing process was 66.7%. In the predictive testing group, 39.5% of individuals tested positive for HD, and in the diagnostic testing group 53%

of individuals tested positive for HD. Nine prenatal tests were requested by five different couples, and 7 tests were performed. Three of these fetuses tested positive for HD (including a set of twins) and these two pregnancies were terminated.

Overall, there seems to be a lack of awareness of and/or access to the genetic services offered for HD through the Division of Human Genetics, National Health Laboratory Service and University of the Witwatersrand, Johannesburg, particularly among black individuals and the professionals treating them. Information generated from this study can be used to understand the individuals seeking genetic counselling and/or testing for HD better, and can direct efforts to improve awareness and access amongst groups noted to be under-represented. It also serves as a starting point for further research.