The Australia and New Zealand Fragile X Family Survey

Introduction

The Fragile X Association of Australia conducted the first national survey of fragile X families in Australia and New Zealand in 2009. The study replicates a similar survey completed in the United States in 2008. The survey form was available online at the Research Triangle International (RTI), North Carolina. Invitations to participate were distributed through the FXAA newsletter, hospital data bases and partner organisations. A trained interviewer was available for those who did not wish to complete the form themselves. The data were collated by staff at the RTI. They are presently being analysed by the project team and a number of papers are in preparation. Caution needs to be exercised in interpreting the results as some cells are quite small.

In this newsletter we present a preliminary overview of the demographics of the sample and an overview of attitudes towards testing for FXADs. Two more overviews will be provided in subsequent newsletters: one on education and employment and the other on availability and quality of services. More detailed analysis will be placed on the website in due course.

Terminology

Fragile X is a group of associated genetic disorders, Fragile X-associated Disorders (FXDs), that affect individuals across generations. Fragile X-associated disorders (FXDs) include:

Fragile X Syndrome (FXS) - most common cause of inherited intellectual disability, behavioural disorders and speech and language delays that manifests in early childhood in males and females;

Fragile X-associated tremor/ataxia syndrome (FXTAS) - neurological disorder which may set in at 50 or over in both males and females, causing tremors, balance and memory problems, and cognitive decline;

Fragile X-associated primary ovarian insufficiency (FXPOI) - causes irregular menstrual cycles, infertility and premature menopause in females.

Demographics of the sample

A total of 113 households representing 289 children responded to the survey. Sixteen households were from New Zealand and 97 from Australia. Of the Australian respondents, 43 were from NSW, 28 from Victoria and 25 from other States. The data presented below include both Australian and New Zealand respondents.

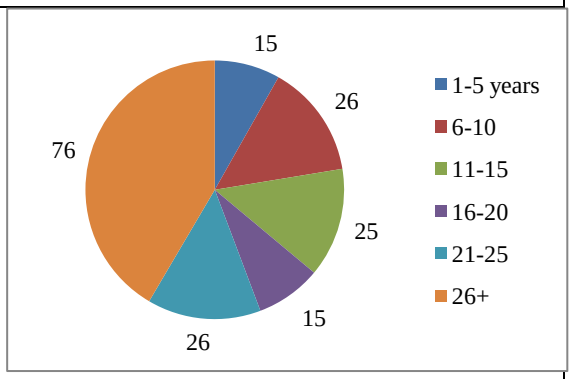
Of the 289 children covered by the survey, 183 were affected by fragile X . Table 1 shows the sex and FXS status of this group of children.

Most (78%) of the 183 lived at home. As Figure 1 shows most full mutation FXS offspring are mature adults, indicating the nature of the long-term commitment of families, particularly to those with the full mutation. One third of all families had a family member who had turned down a job because of their FXS commitment. Forty nine families (nearly half) indicated their child needed either moderate or considerable assistance in day-to-day living.

Table 1: Status of Fragile X among the children

Fragile X Status of Child	No.	%
pre-mutation male	13	7.1
pre-mutation female	18	9.8
full-mutation male	111	60.7
full-mutation female	41	22.4
Total	183	100.

Figure !: Age of children with full mutation



Occurrence of co-occurring conditions by sex and mutation status

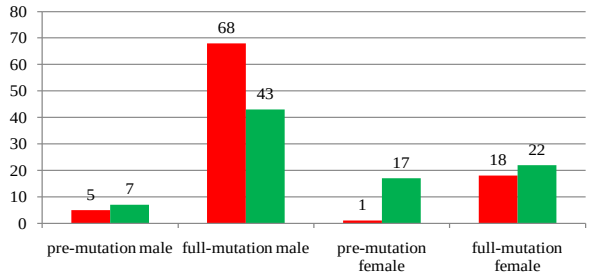
The following charts show the variability of co-occurring disorders — problems of attention, hyperactivity, aggression, self-injury, autism, seizures, anxiety and depression. For full mutation individuals, co-occurring conditions were quite similar though males have relatively more problems with attention, hyperactivity, aggression, self injury and anxiety.

One of the important findings of the study is this difference in co-occurring conditions as experienced by *pre*-mutation FX males and females. At the pre-mutation level it is clear that while females have less difficulty with attention, hyperactivity or aggression they are considerably more prone to depression and anxiety. These differences carry many implications for education and behaviour management.

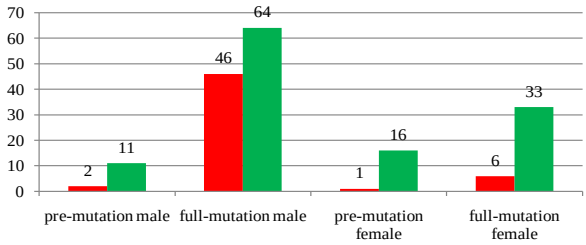
Charts of co-occurring conditions (number of persons)

=Yes = No

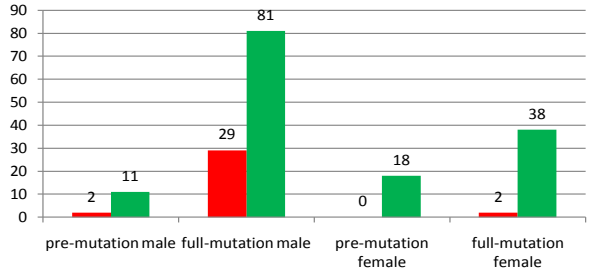
Diagnosed with attention problems



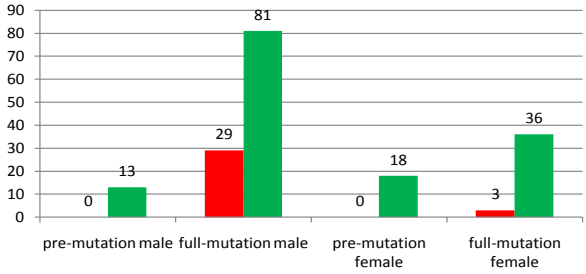
Diagnosed with hyperactive problem



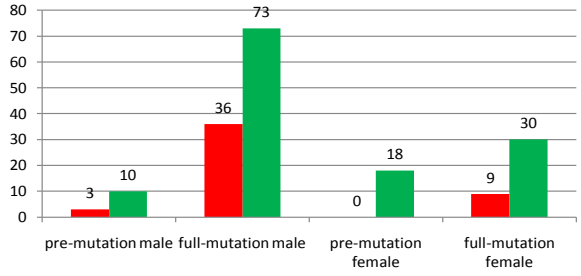
Diagnosed with aggression problems



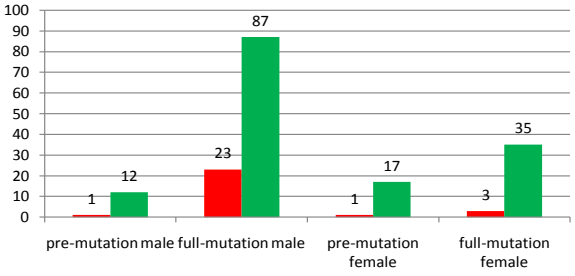
Diagnosed with Self injury problems



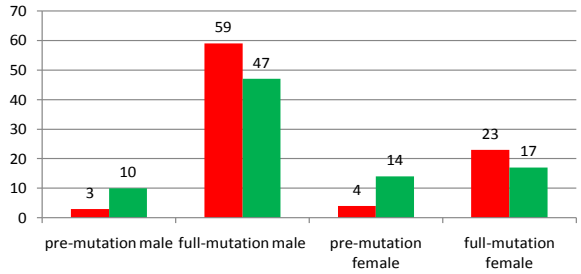
Diagnosed with autism problems



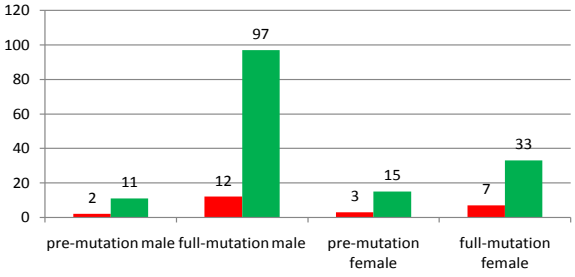
Diagnosed with seizures problem



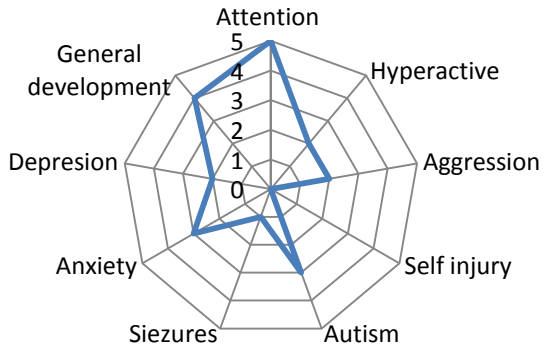
Diagnosed with anxiety problems



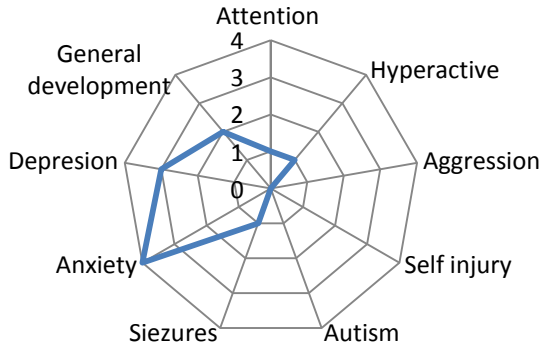
Diagnosed with depression problems



Pre-mutation Males (n=13)



Pre-mutation Females (n=18)



Knowledge of FXDs and attitudes toward testing

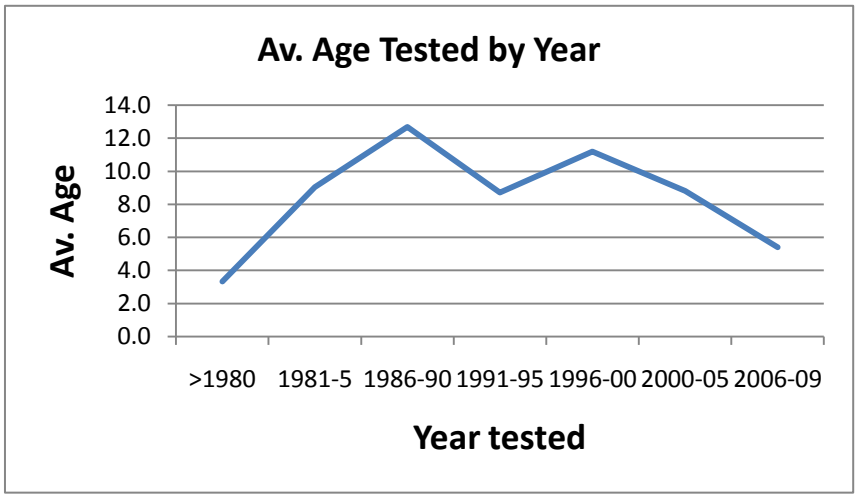
The sample data show that only 12 out of 113 respondents knew about their own or their partner's FX carrier status before their first affected child was tested. The age of testing for FXDs has changed over time. Through the 1980s, there was an increase in the average age at which a child was diagnosed. No doubt this reflected a new awareness of FX syndrome and the technological capacity to test. More recently the average age has dropped to under 6, but this is still quite high compared to similar data from the US.

A number of questions were asked to gauge attitudes to testing for FXDs at various stages:

81% of respondents indicated that the best time to offer testing for both men and women is before a woman gets pregnant.

If pre-pregnancy testing to determine carriers has not been done, most parents (how many?) would prefer to know, during pregnancy, if their child was affected. A small number of respondents (9) indicated they did not wish to know during pregnancy;

Post-natal or newborn screening of all babies was even more strongly supported by % of respondents.



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