Females Who Have Fragile X Syndrome

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Early Work:

- Miezejeski et al., 1986
  - Difficulties with arithmetic, digit span & block design
- Freund & Reiss, 1991
  - Poor at short-term memory for non-verbal sequential material
  - Good at verbal short-term memory for sequenced information
- Brainard et al., 1991
  - Premutation carriers relatively unaffected
  - ? Slight weakness in verbal short-term memory
Mazzocco et al., 1992:

- Cytogenetically expressing individuals
  - perseveration
  - tangential speech
  - impulsiveness
  - distractibility
  - difficulty with transitions
  - problem-solving difficulties - especially with need to consider additional information simultaneously

- Perspective-taking & emotion perception O.K.
Also....

- Same uneven cognitive profile as in boys, even in those with at least average IQ (Cronister et al., 1991)

- Fragile X syndrome is as common in girls with autistic spectrum disorders as in similarly affected boys (Cohen et al., 1989)
Pilot Study on Young Female Carriers

- 10 individuals aged 5-20 years
- 5 full mutation, 5 premutation

Assessments:
- psychological testing of intellectual functioning
- childhood behavior checklist
- autistic spectrum disorder screen
- child depression inventory (where possible)
- worry scale (where possible)
Intellectual Functioning:

- **Full scale IQ:**
  - average 58, range 40-80
  - lower IQ’s usually but not always associated with full mutations

- **Verbal IQ:**
  - average 60, range 31-87

- **Performance IQ:**
  - average 60, range 46-78

- No consistent trend for verbal or performance skills to be stronger
Social & Language Functioning:

- **Autistic Spectrum Disorder in 5**
  - 3 premutations, 2 full mutations
- **Irrespective of diagnostic label:**
  - shyness, social anxiety, self-consciousness, easily embarrassed, social isolation
  - obsessional including obsessive worrying
  - problems socialising
  - Social use of language & “semantic/pragmatic problems
Concentration Skills:

- 5 fulfil criteria for Attention Deficit-Hyperactivity Disorder
  - 3 full mutations, 2 premutations
- Irrespective of diagnostic label:
  - inattentiveness, restlessness, impulsiveness, distractibility, “wandering mind syndrome”
- No evidence of any tendency towards clinical depression. But low self-esteem problematic
Other Emotional & Behavioural Difficulties

- Clingy & dependent
- Fearful
- Executive Function Problems
  - planning & organising thoughts
  - shifting topic of thought
  - problem-solving
  - perseveration
  - difficulty focussing the mind
Conclusions:

- As with boys, girls can show a wide range of affectedness, even with a premutation.
- Autistic spectrum & attention deficit disorders are common even with reasonable intellectual functioning.
- Low self-esteem is common but avoidable.
- Shyness, social anxiety, difficulty socialising and language problems are common.
And The Good News!

- All the above can be helped!
- Obsessional tendencies offset impulsivity & difficulties focussing mentally.
- Excellent in helping to adhere to treatment regimes.
- May be problematic if starts to interfere with daily living.
Problems experienced by adult females who have fragile X syndrome

- Uneven intellectual profile irrespective of IQ
- Features of ADHD, mental disorganisation & executive dysfunction
- Non autistic social impairments
  - Shyness, social sensitivity & social anxiety
  - Oversociability, garrulousness & overintrusiveness
  - Tendency towards impulsive social faux pas
  - Perseverative social, language & behavioural styles
  - Impressionability & suggestibility → vulnerability
  - May → attractiveness or negative impression
- Ruminative anxious depression