



Fragile X Syndrome and Autism: similar but different

A parent's view

Dr Lucia Elghali
Director, The Fragile X Society

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Why is this important?

- **Fragile X Syndrome and Autism**
 - Relationship between conditions is contested
 - Is a co-morbid diagnosis of Autism helpful? To whom?
- **What is the impact for us as a family?**
 - Diagnosis
 - Support services offered (education and health)
- **Service provision: personal reflections**
 - What to consider in “Early Years” provision
 - Read across to other “Rare Diseases”?



Autism and FXS: a lay view

Autism

- **Diverse causes** (genetic, environmental)
- “Catch all” or “umbrella” diagnosis for a cluster of similar looking **behaviours**
- **Very heterogeneous expression**

Fragile X Syndrome

- A specific **genetic diagnosis** caused by **>200 CGG repeats** at **FMR1 gene locus** (“methylation”)
- Expression varies, though has a **distinct profile**
- **Physical and behavioural**



FXS and Autism

- Autistic-like behaviour is part of FXS profile e.g.
 - Gaze avoidance
 - Hand flapping
 - Many may also meet diagnostic criteria for autism
 - DSM IV or ICD 10 still used in NHS Wales
 - “Triad of impairments”
- **FXS Profile distinct from Non-Syndromic Autism**
- E.g. Hall *et al* (2010) “category mistake”



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Why does this matter? (1)

- Autism Diagnosis (with ADI-R and ADOS)
 - Detects Idiopathic or Non-Syndromic Autism
 - Known problems with FXS: not sensitive enough
 - E.g. Gaze avoidance and social anxiety
- Diagnosing Autism in FXS Individuals
 - Why necessary? Cause and profile already known
 - Autism diagnosis should not be used as a measure of severity of FXS traits: qualitatively different



Why does this matter? (2)

- Our experiences with provision of services
 - Many are “diagnosis led”: Autism diagnosis to access
 - Interventions may help **if adapted with care**
- FXS and Early Years: some examples
 - “Portage” offered by NHS
 - Interventions based on the science of Applied Behaviour Analysis (e.g. EIBI) obtained privately at USW
 - “Promoting independence” by changing 1:1 TAs?

→ ONE SIZE DOES NOT FIT ALL



Wider reflections...

- Don't forget about unaffected siblings/family
- Look at the child, not the diagnosis
 - Just because it looks like “autism”, doesn't mean children with FXS respond in the same way
- Access to services independent of diagnosis
 - Conflicts with plans for Autism Specific legislation?
- Rare Diseases Strategy: FXS specialist centre?
 - Complex issues with diagnosis and interventions
 - Advice and referrals
 - Similar issues with other rare conditions?



Thank You!

Some key references on FXS and Autism

- Abbeduto, L., McDuffie, A., & Thurman, A. J. (2014). *The fragile X syndrome–autism comorbidity: what do we really know?* *Frontiers in Genetics*, 5, 355.
- Hall, S. S., Lightbody, A. A., Hirt, M., Rezvani, A., & Reiss, A. L. (2010). *Autism in Fragile X Syndrome: A Category Mistake?* *Journal of the American Academy of Child and Adolescent Psychiatry*, 49(9), 921–933.
- McDuffie A., Thurman A.J., Hagerman R.J. & Abbeduto, L. (2015). *Symptoms of Autism in Males with Fragile X Syndrome: A Comparison to Nonsyndromic ASD Using Current ADI-R Scores.* *Journal of Autism Developmental Disorders*, 45(7), 1925-37.