

Females with Fragile X Syndrome

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Females with Fragile X Syndrome

- ▶ What I know about Females with FXS- They work hard to overcome their deficits
- ▶ What I have learned from them- They persevere and no matter how high the mountain, they continue to climb with determination. When they become discouraged, they rewind and move forward
- ▶ How they inspire me- They continue to laugh and enjoy life even when they struggle- they are my heroes!

Females FM – cognitive profile

- ▶ IQ – 1 SD < N , 70% < 85, 50% < 70,
- ▶ Lower arithmetic score, similarities, EF, spatial-visual
- ▶ Higher verbal scores, picture completion, comprehension, symbol search

Striking EF deficit, spared verbal skills

(Bennetto '01 – N & IQ matched)

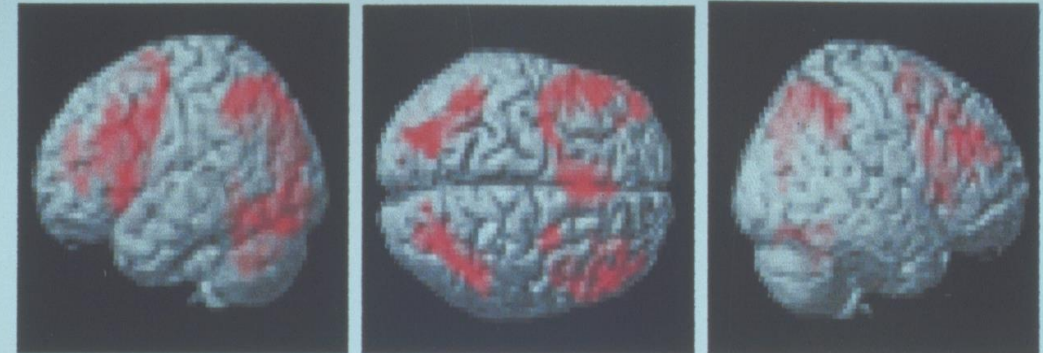
Functional Studies

Decreased activation

- executive function (Menon et al 2000)
- Visuospatial processing (Kwon et al 2001)
- Math tasks (Rivera et al 2002)

Fronto-parietal-cerebellar

Control Group Activation-Math Task



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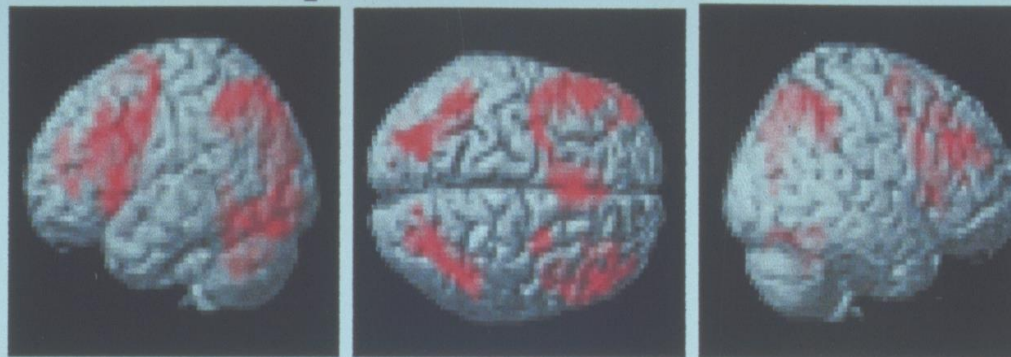
Fronto-parietal-cerebellar

The extended neural network cannot be recruited to solve more difficult tasks

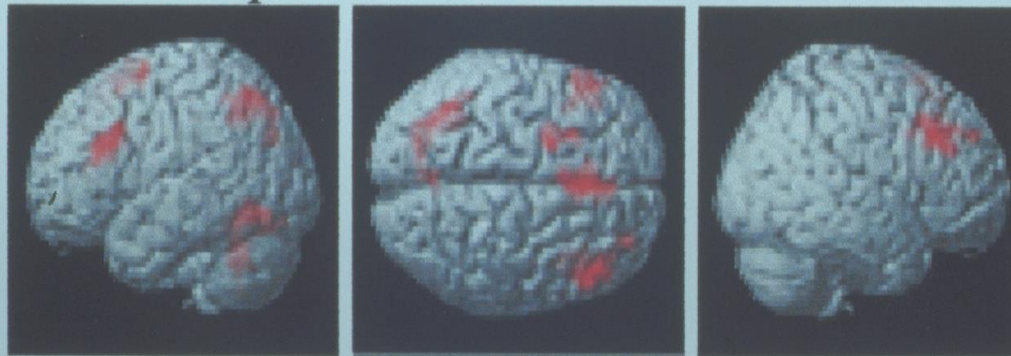
These deficits correlate inversely with FMRP

The more FMRP the better the activation

Control Group Activation-Math Task



FraX Group Activation-Math Task



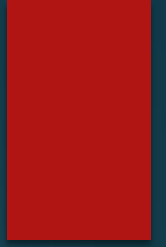
Females FM - behavioural profile

- ▶ ADHD - 35% (Hagerman 2000)
- ▶ Shyness, social anxiety common
(Mazzocco 1998, Sobesky 1995)
- ▶ Anxiety - 40% (Lachiewicz 1994)
- ▶ Avoidant personality - majority (Freund 1993)
- ▶ Selective mutism (Hagerman 2000)

Females

- ▶ Variance in functioning is not correlated with number of CGG repeats
- ▶ Some females with a full mutation function in the normal range
- ▶ Some pre-mutation females have learning disabilities and can manifest emotional difficulties as well

Females who pass on the gene have a significant connection with their children and can be an invaluable resource to gaining insight and understanding



Females

- ▶ Cognitive Profile in Females with FXS
 - ▶ Not enough consistent evidence to determine decline in IQ
 - ▶ Generally, Performance IQ < Verbal IQ
 - ▶ Lower scores on Block Design, Arithmetic and Digit Span
 - ▶ Difficulty with abstract verbal reasoning (Similarities)
 - ▶ Weaknesses in short-term auditory memory and visual spatial ability

Females

- ▶ Spatial Ability in Females with FXS
 - ▶ Varied opinions depending on research bodies
 - ▶ Weaknesses in visual spatial logic (Cianchetti et al 1991)
 - ▶ Weaknesses in figural memory as opposed to verbal memory (Mazzocco et al 1993)
 - ▶ Deficits in spatial short-term memory (Kemper et al 1986)

Females

- ▶ Major Learning Issues for Females with FXS
 - ▶ Information Processing Differences
 - ▶ Stronger Verbal memory (Mazzocco et al 1992)
 - ▶ Poorer Performance on Digit Span, Verbal Learning and Working Memory
 - ▶ Deficits in Spatial Memory, Motor Coordination and Arithmetic (Mazzocco et al 1993)

Females

- ▶ Executive Function in Full Mutation
 - ▶ In comparison studies using various controls, performance of women with FXS, the most consistent difference was in EF
 - ▶ EF deficits seem consistently low across ages: present in both childhood and adulthood
 - ▶ Pragmatics, organizational aspects of memory, behavioral issues such as hyperactivity or impulsivity may contribute to poor EF

Females

- ▶ Executive Function in Females with FXS
- ▶ Strategic Planning and Sequential Problem Solving
- ▶ Impulse Control or Selective Attention
- ▶ Flexibility of Thought and Action

Females

Behavioral Characteristics

- Shyness
- Superficially extroverted
- Poor impulse control
- Lack of personal boundaries
- Non-verbal learning disabilities
- Conflict avoidance

Psychiatric Conditions

- Depression
- Anxiety-social phobias
- Obsessive- compulsive personality
- Bipolar disorder
- Dependent personality

Problems in Activities of Daily Living

- ▶ Money management
 - ▶ Issues with math makes managing money difficult. They often over spend, do not understand the value of money and lack an understanding of what things cost
 - ▶ They can obsess over collections of items of high interest such a sports, pop music and movie memorabilia

Problems in Activities of Daily Living

- ▶ Organizational Skills
 - ▶ Females may have difficulty keeping things organized, including personal items and home management
 - ▶ Clutter can accumulate making it difficult to find things

Problems in Activities of Daily Living

- ▶ Relationships are difficult
 - ▶ Females can be used by controlling males
 - ▶ Females are often unable to express their true feelings due to aversion to conflict
 - ▶ Females may become angry and abusive in a relationship when they feel safe

Problems in Activities of Daily Living

- ▶ Work Relationships can be difficult
 - ▶ Often too stressful to work 8 hr days
 - ▶ If the work environment includes social expectations, it is more difficult
 - ▶ If there is a time limit to turn out a product, it can be difficult
 - ▶ Interpersonal relationships are problematic when there is conflict or disagreement

Vocational

- ▶ Females may enjoy working with children- many young girls choose the path of early intervention
- ▶ Some females choose to work with animals at a shelter or veterinarian's offices
- ▶ Females may enjoy food preparation or working for a catering company if the job does not require fast food pace
- ▶ Females are competent in using eBay, posting collections to be sold on the internet or writing movie blogs

Case Study #1

24 year old

FXS Full mutation

Diagnosed at the age of 7

My Interests:



FXS affects my life:

- Anxiety is my major affect of FXS
 - meeting new people
 - new situations
 - school
 - learning to drive
 - having to rush/being on time
 - problems sleeping
 - I feel angry and act angry to others
 - having too much to do in a day

What's going on now:

- Working at Aunt Kathy's Childcare with Toddlers(Mostly with one year olds)
- Working with Dr.Erickson on Medications for Anxiety.
- Currently attending Gateway to get an Associates Degree in Education. Also attending NKU.
- Learning to Drive

Future Plans



- Continue to work on Celebration Retreats
- Continue to work at Aunt Kathy's Childcare
- Get my Driver's License
- Get my Associates Degree
- Get my Bachelors Degree at NKU



Case Study #2

AGE 21

FULL MUTATION

About Me

- ▶ I was diagnosed with FXS when I was 9 after my brother was diagnosed at age 4. He is more affected than I am.
- ▶ I have another family member who has FXS
- ▶ I am getting a BA in psychology next year
- ▶ I want to become a counselor for kids with disabilities like autism and FXS
- ▶ In my spare time I like to kayak, hike, and be with my friends.
- ▶ I love working with teens with disabilities
at my job with the Therapeutic Rec Program.

My Struggles

- ▶ My biggest struggle was the barrier between teacher and student relationship. Mostly revolving around the subject math.
 - ▶ Example one: Assuming I was lazy and not taking the time to help me.
 - ▶ Example two: How I solved this problem through accommodations.

My Struggles

- ▶ My biggest challenge is anxiety
- ▶ I had a big drive to be a chef so I took a job at a four star restaurant.
- ▶ The pressure there was too stressful because it was a top rated restaurant. If I messed up I got yelled at.
- ▶ I had to make 10 of these tuna towers in less than 10 minutes



My Successes


- ▶ I made it into the National Honor Society in High School. In order to qualify I needed a 3.0 GPA but I had a 3.5GPA. I had to be tutored in math
- ▶ I was number one on my team on the butterfly stroke.

Advice to Parents

- ▶ Always spend time with your child. Listen to them and try to understand their struggles.
- ▶ Support them through the good times and bad times
- ▶ Always remind them that even though they have FXS, it does not limit or define your character

Advice to Teachers

- ▶ Always, always show patience
- ▶ When teachers get upset or yell, it feels like there is a spotlight on the person with FXS. It feels like everyone is looking at you and your disability is being exposed
- ▶ Work at a pace that allows for success- we need time to process and learn
- ▶ Show encouragement- do not show frustration
- ▶ Also make time to provide the appropriate accommodations.
 - ▶ For example read the tests out loud or let them use their iPod during math quizzes or tests as I have advocated for these things.



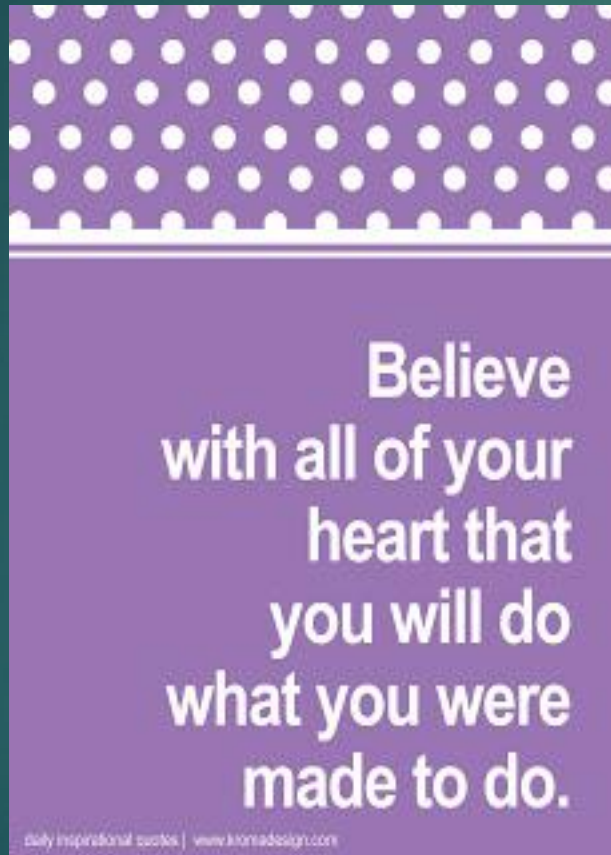
Case Study #4

21 years old

Background

- ▶ I was 15 years old when I was diagnosed
- ▶ One of my mom's relatives died and his DNA was positive for FXS
- ▶ I was tested because I had always struggled at school
- ▶ I have one brother who does not have FXS

My Strengths



- ▶ This is one of my favorite quotes
- ▶ I felt I was made to work with children
- ▶ I am attending Pikes Peak Community College
- ▶ I am working on an associate's degree in Early Childhood Development

My Strengths

- ▶ I am strong; I am able to do things I never thought possible
- ▶ I am positive; I always try to find the silver lining
- ▶ I am happy; I love to laugh (sometimes it is a nervous laugh)
- ▶ I am determined; I never give up

My weaknesses

- ▶ Socially, I am typically shy. It is hard for me to introduce myself to others. I often close my eyes or look away during a conversation.
- ▶ Sometimes I get too dramatic about personal things which can put me in difficult social situations.
- ▶ School is difficult, especially math
- ▶ I had some difficulty in my college practicum last year with my mentor teacher. She expected me to “get over” having FXS. This was frustrating because it was clear she did not understand my challenges even after I explained it
- ▶ I learned to how to advocate for myself during this encounter

What I'm doing this year!

- ▶ I'm working at the Picnic Basket Catering company. I work in the delivery department.
- ▶ I am going to be starting my junior year of college this Fall.
- ▶ I am playing tennis this summer which is something that I absolutely love.



How I feel about having FXS

- ▶ Having FXS has impacted my life so much but I wouldn't change it because I've been able to inspire so many of my friends and family with my story.
- ▶ I felt very alone when I was first diagnosed with FXS because I didn't know much about it but once I learned more about it I began to accept the fact that I have FXS.
- ▶ My message for you is to never give up and that you are not alone in this!!! One of my favorite quotes is:"you got this!"

My Accomplishments



- ▶ I graduated from High School in May of 2014
- ▶ I finished high school with a GPA of 3.50!
- ▶ I am now in college which is something that I never thought possible.
- ▶ I recently finished my sophomore year of college with straight A`s!

Advice to Parents

- ▶ Stay positive
- ▶ Don't yell when we do something wrong; explain what was wrong and then try to help fix it
- ▶ Encourage social participation, but don't push it
- ▶ Treat your child as if they were normal but allow for some special support when needed.



Questions?