A Day of Best Practice

• Today you are in for a treat!
• We wanted to have a full day to share “Best Practices” across disciplines and developmental areas to help guide you in advocating for best services for individuals with FXS across the lifespan
• All of us are passionate and know that we can provide proper care, support, and advocacy for individuals with FXS based on what we know about FX specifically
• We all support a multidisciplinary team approach, with the family fully a part of the team; the team must work in collaboration/cooperation with each other - this is best practice in every field!

From X in the genes to X on the treatment map!

Phenotype: What we know from the science about the gene impact on brain, fx, and learning/behavior
FX Presentation related to how Occupational Therapists provide supports
FX Phenotype specifies these needs for the individual
Interventions – always individualized and modified for FX
Fragile X Phenotype

• Affects about 1:4000 males, 1:6000 females

• Neuro Phenotype (VARIABLE)
  - Mild/Moderate ID
  - Better adaptive function than IQ
  - Hypersensory and Sensory difficulties
  - ADHD
  - Autism or ASD like-symptoms
  - Social Approach/Withdrawal
  - EF difficulties
  - Motor Difficulties
  - Aggressive Behavior
  - Seizures

• Non-Neuro Phenotype
  - Facial characteristics (non-diagnostic)
  - Connective tissue abnormalities (joint laxity, mitral valve prolapse)
  - Strabismus, Recurrent OM, GER


Phenotype of FX related to domains treated by Occupational Therapists

• Characteristics

  Cog  Soc  Sens  Motor  Language  Adaptive  Imitation  Gestalt

• How OT addresses them

The "Frag-i-cus"

What is Occupational Therapy?

The term “occupation” comes from how we “occupy” our time. Our daily life’s roles and activities including self-care, play, work, social engagement, leisure, and learning are all daily “occupations”.

• Sensory integration differences often greatly impact one’s fulfillment of daily occupations.
**What is Occupational Therapy?**

![Diagram of Occupational Therapy]

Current “best practice” in field of occupational therapy includes use of: “P-O-E”, “Sensory-Based”, “Relationship-Based”, “Play-Based” models that are grounded in neuroscience and developmental science (i.e., “Brain-Based”):

- Use purposeful and meaningful activities to maximize potential
- Intervention is evidence-informed and guided by ongoing monitoring of intervention plan, effectiveness, and how needs are targeted (data driven, but relationship based)
- Intervention at TWO LEVELS – differentiates from other therapies:
  - Work on “person” – factors that impair function as well as
  - to improve adaptive daily occupational functioning.

---

**Occupational Therapy**

- Across ages and stages, individuals with FXS may benefit from OT services (El/School/Facility/Private)
- Direct Treatment
  - When skills need to support to develop (1:1, grp, home Programs)
  - When environments, people in those environments, the demands/tasks at hand, or the individual is not operating without stress (distress)
- Consultation (Indirect intervention)
  - To navigate rocky patches along the road of life
  - When current team needs specialized support
FX Phenotype/ OT Areas: Areas often requiring treatment

- Hypersensitivity to visual, auditory, olfactory and tactile stimuli
- Hyperarousal and anxiety
- Difficulty with motor planning and sequencing
- Perseveration
- Attention difficulties
- Hyperactivity and Impulsivity
- Cognitive deficits including EF
- Social skills difficulties
- Life, work, school skills difficult

The "Frag-i-cus"

Brain is complex, more than just cognition or stimulus → response

Neural Sensory Pathways (SI/SPD)

Sensory Discrimination
Postural Development
Sensory Modulation
What is Sensory Integration (SI)?

- Sensory Integration (SI) is defined by Ayres as: *The organization of sensation for use.*
- SI issues also known as sensory processing differences impact:
  - Attention and focus, Behavior, Language processing
  - Learning, Overall functioning
- Research has well documented SI issues in many individuals with FXS
- These issues require specialized intervention that matches the FX phenotype
- SI Intervention helps with core issues seen in FX

Sensory Integration contributions to Fragile X Characteristics

<table>
<thead>
<tr>
<th>Characteristic of FraX</th>
<th>Possible SI/S-A-M</th>
</tr>
</thead>
<tbody>
<tr>
<td>hypersensitivity to stimuli</td>
<td>X</td>
</tr>
<tr>
<td>poor eye contact</td>
<td>X</td>
</tr>
<tr>
<td>attentional system difficulties</td>
<td>X</td>
</tr>
<tr>
<td>hyperactivity / impulsivity</td>
<td>X</td>
</tr>
<tr>
<td>difficulties with motor planning and sequencing</td>
<td>X</td>
</tr>
<tr>
<td>hand flapping and biting</td>
<td>X</td>
</tr>
<tr>
<td>hyperarousal and anxiety</td>
<td>X</td>
</tr>
<tr>
<td>problems with transitions</td>
<td>X</td>
</tr>
<tr>
<td>perseveration</td>
<td>X</td>
</tr>
<tr>
<td>social interaction challenges</td>
<td>X</td>
</tr>
<tr>
<td>life, work, school difficulties</td>
<td>X</td>
</tr>
<tr>
<td>cognitive difficulties including EF</td>
<td>X</td>
</tr>
</tbody>
</table>

Brain Based Approach to Understanding the Role of OT in FXS

Dan Siegel’s handy brain anatomy model
• Hyperarousal & Anxiety
  − Primary issue, across lifespan
  − While hyperarousal can produce ‘behaviors’, they are a result of the biology, not learned per se.
  − As such, Tx requires brain-based approach, not behavioral approach

• Brain Based Approach to treating, managing, living with hyperarousal

Brain Based Approach to Treating, Managing, Living with Hyperarousal

“Please, stop! Help me feel safe and ready!”

Treatment comes from a base of safety and comfort, within an attuned relationship, to promote engagement. This results in improved capacities and skills and a cycle that builds forward...

Proactive Management of Hyperarousal
Neurologically, we have to address the low-route systems in relational context

OT Intervention
Best Practices for supporting hyperarousal and anxiety

- Goal to minimize hyperarousal and anxiety and increase coping/self-regulation
- OT’s start from the “bottom up” in treating these issues
- SI theory and practice assist
- Sensory Diet and Self-Regulation are the focus
- We incorporate higher level skills, only AFTER establishing the base!
- Come to the sensory diets/self-regulation talk for more

Components of the S.T.E.P.S.I. Model
Used in OT to create comprehensive intervention

S ensation
T ask
E nvironment
P redictability
S elf-Regulation
I nteraction

Clinical/Reasoning and Fidelity to Treatment Model; Stackhouse, Wilbarger, Trunnell, (1998)
FX & OT
PERSON

- Low muscle tone, loose connective tissue impacts sensory motor skills, posture, motor planning
- Overall skill acquisition

OT Approach to treating, managing, living with difficulties

OT Intervention
Best Practices for supporting motor based skills

- Build strength and ensure alignment to compensate for low muscle tone
- Remember IMITATION and GESTALT learning strengths in FXS, don’t teach parts, teach whole
- SI theory and practice assist
- Interest areas and motivation matter!
- Meaningful repetition needed for motor learning
Posture and Praxis

Hands as Tools, as an expression of self, and an in-road to the world

Oral Motor, Feeding, and Respiration
FX & OT

- Autism within FXS
- Imitation and Motor Planning
- Skill Learning
  - FM
  - Safety and Environments
  - Independence, ADL's
  - Meaningful life engagement
- Brain Based Approach to treating these concerns

Autism in Fragile X

The Social Approach Scale paradigm identifies dynamics of social avoidance and indifference that can distinguish Autism from Social Anxiety in Fragile X

S-A-M Functions Related to Social Cognitive Behavior

- Shares and understands emotional warmth with others
- Adapts to daily routine
- Engages with others - intersubjectivity
- Adapts to changes
- Settles and soothes - eventually self calms – self-regulates arousal and emotional state
- Uses social motivation adaptively

* High baseline and post-stimulus cortisol levels are seen in FXS-ASD
Roberts et al. JADD 37:1748-60, 2007
OT Intervention

best Practices for supporting social and autism related concerns

• Goal to improve social connection, this drives the rest in FXS!
• OT’s start from a base of regulation, connection, and build a sense of competence

• Anxiety makes those w/ FXS look more ASD-like – address anxiety to know what is really afoot!

DIR/and ESDM models need to be OT informed
Other ABA models need to be modified, require OT input

Role as Student and Classmate

Play Skills in FXS
Balancing Daily Engagement

Daily Living and Self-Skills

Honor the person for who they are.
Provide OT when needed, it will make a difference in daily life!
OT can help across ages and stages and is an integral part of your FX team!
OT is at the base....next, behavior! Have a great day of best practices!!

Phenotype: What we know from the science about the gene impact on brain function and learning/behavior

FX Presentation: related to how Occupational Therapists provide supports

FX Phenotype specifies these needs

Interventions

Thank YOU