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NATIONAL **FRAGILE X** FOUNDATION
FRAGILE X CLINICAL & RESEARCH CONSORTIUM

Consensus of the Fragile X Clinical & Research Consortium

AN INTRODUCTION TO ASSESSING CHILDREN WITH FRAGILE X SYNDROME

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Readers of this document who wish to learn more about specific assessment measures and tools, including their strengths and weaknesses, may wish to review the document ***Assessment of Individuals with Fragile X Syndrome: Detailed Recommendations for Clinicians, Providers and Other Professionals*** which also includes a list of references.

GLOSSARY

Assessment/Assessment tool: Tests, questionnaires, interviews, etc. to measure a person's skills.

Assessor or Examiner: The person conducting the assessment.

Cognitive flexibility: The brain's ability to transition from thinking about one concept to another.

Lability: Liable to change; easily altered, as in rapid changes in mood.

Level of Development or Functioning: Assessments are used to determine cognition, communication, sensory processing abilities and motor skills.

Maladaptive: Behaviors that are potentially harmful to self or others; prevents someone from making behavior adjustments that are in their own best interest. (e.g., verbal and physical aggression are considered to be maladaptive in most situations.)

Neuromotor testing: Evaluates motor functions through the age at which a skill is expected to be mastered.

Norm-referenced test/Standardized measure: designed to compare and rank test takers in relation to others of the same age (established by prior testing of large sample of typically developing individuals at various ages). Also, a form of test that requires all test takers to answer the same questions, and that is scored in a "standard" or consistent manner, making it possible to compare the relative performance of individual students or groups of students.

Phenotype: A set of observable characteristics or traits of an individual resulting from the influences of genetics and the environment.

Psychometric test: Quantitative tests which give numeric scores or age equivalents for the measurement of psychological variables such as intelligence, aptitude, and personality traits.

Psychometric standards: A standard and scientific method used to measure individuals' mental capabilities and behavioral style.

Reliability: The extent to which test scores are consistent across different occasions of testing, different editions of the test, or different raters scoring the test taker's responses.

Response inhibition: Refers to the ability to suppress responses or actions that are not appropriate or no longer adaptive to a situation.

Self-harm/injurious behaviors: Behaviors that cause physical harm to the individual (examples include hand biting or head banging).

Side conversations: Strategy to minimize anxiety and hyperarousal in an individual with FXS by minimizing direct communication.

Stimming: Self-stimulatory, repetitive motor behaviors that serve a purpose or provide positive internal feedback for the individual. Examples include hand-flapping and rocking.

Response to intervention (RTI): A process used by educators to help students who are struggling with a skill or lesson; every teacher will use interventions (a set of teaching procedures) with a student to help them succeed in the classroom and should use quantitative (numeric) measurement to determine whether the student is responding to the intervention appropriately by making progress.

Validity: The extent to which a test accurately measures what it is supposed to measure.

Working memory: A form of memory that allows a person to temporarily hold a limited amount of information in his or her mind to have it ready for immediate mental use. It is considered essential for learning.

OVERVIEW

Assessment of individuals with delayed development is important for many reasons. Foremost among those reasons is to provide the family, and the educational, medical and therapeutic team around them, clarity in identifying strengths and needs and aligning these to supports and interventions. Multidisciplinary team assessments are suggested to appropriately design educational intervention programs that address the individual's needs starting at the level where they are currently performing. (i.e., "level of functioning" or "developmental level.") Other important reasons for assessing children with developmental delays is to recognize any delays as early as possible to allow for early implementation of educational interventions and to create a baseline so that future assessments can show if progress is being made and to inform adjustments to ensure best outcomes.

Assessment of individuals with fragile X syndrome (FXS) is challenging in numerous ways, ranging from choice and limitations of instruments to behavioral and emotional factors in the individual that may impact the testing process, to scoring and interpretation. Fortunately, decades of research and clinical experience pertaining to assessment, including recent detailed studies of the performance of several measures as outcome measures for clinical trials, have provided very useful guidance.

This document will cover several important general considerations for the clinical assessment of people with FXS, including:

- Fragile X Syndrome Phenotype
- Principles of Assessment Selection
- Preparation and Approach to Testing
- Accommodations that can be Made to the Testing Process

We will also cover the primary domains of assessment, including recommended assessments:

- Cognition
- Behavior
- Social and Emotional Functioning
- Academic Skills
- Speech/Language/Communication
- Neuromotor Functioning and General Motor Functioning
- Sensory Processing and Adaptive Functioning

with additional sections on assessment of infants and toddlers and autism.

Fragile X Syndrome Phenotype

A phenotype is a set of observable characteristics or traits of an individual resulting from the influences of genetics and the environment. All assessors should have an intimate understanding and awareness of the cognitive, behavioral, social-emotional academic, speech/language, and neuromotor phenotypes of FXS. This ensures that they will choose appropriate assessment tools, administer the assessment in ways that are sensitive to the common challenges encountered, recognize critical strengths and weaknesses when they are present, and interpret and report results in a manner that are both meaningful and accurate.

The FXS cognitive phenotype may include particular difficulties with sequential processing of information, attention, working memory, response inhibition, cognitive flexibility, and math.

The FXS behavioral phenotype is often characterized by hyperactivity and impulsivity, inattentive behaviors, repetitive or stereotyped behavior and speech (which may be characterized as features of autism), self-injury and irritable/aggressive behavior.

The FXS social-emotional phenotype includes symptoms of anxiety, irritability, mood changes, obsessive or perseverative actions, social approach-avoidance (including gaze avoidance) and social communication delays (which also may be characterized as autism).

The FXS academic phenotype often includes skills that are delayed in all areas, or where there is a deficit, including with reading, writing, and math.

The FXS speech/language/communication phenotype includes delays or disorders in all areas of speech and language, but particular features are rapid speech, cluttering (speech that sounds rapid, unclear and/or disorganized), repetitive or stereotyped speech (which may be characterized as a feature of autism).

The FXS neuromotor phenotype can include hypotonia, delayed fine and gross motor skills, and difficulty with coordination. These early symptoms may be the first presenting signs that lead to FXS testing and diagnosis.

It is important to note the many positive aspects often found in children and adults with FXS including that they can be very social and friendly, have excellent imitation skills and strong visual memory/long term memory. They often like to help others, want to please and make others happy, are nice, thoughtful, and have a wonderful sense of humor.

Principles of Assessment Selection

One of the most important aspects of FXS that must be considered in the assessment process is developmental level. Given that most males and many females function in the intellectual disability (ID) range, knowing a developmental age estimate prior to assessment (often based upon an initial interview with a caregiver and behavioral observations) will greatly aid the assessor in choice of instruments and starting points for tests.

There are several factors to consider when choosing an assessment (i.e., tests, questionnaires, interviews, etc.) for a person with FXS:

- A general rule of thumb is to select assessments that have demonstrated the usual psychometric standards of reliability and validity.

- It is advisable to use assessments that have specifically established reliability and validity in individuals with FXS. This provides added assurance that the tool is more likely to be helpful.
- If these FXS-specific assessments are not available, the next best option is to confirm that the tool is valid for persons with similar disabilities who do not have FXS (e.g., general intellectual disability).
- It is important to note: As there is a range of abilities in FXS (particularly among females or males with mosaic FXS), assessments that are appropriate for those with FXS and ID, may not be appropriate for all individuals with FXS.

Important Note: assessments and measures are developed using typically developing persons: Many, if not most, measures, including behavioral and emotional measures, are developed using typically developing persons. What this means is that an assessment is likely to yield scores that compare the individual with FXS to their age-peers, not to other people of the same developmental level. In addition, some assessments may be inappropriate for use in FXS as they do not accommodate for the behaviors and other characteristics of those with FXS. Lastly, assessments developed for older adolescents and adults, especially, may have materials and items less suited for individuals with FXS with lower mental/developmental ages, thus making it harder for them to engage and participate. ***One may wish to discuss this issue with the assessor in advance of the assessment.***

Assessment Strategies

There are several considerations for administration and scoring of developmental testing for young children with FXS. Children with FXS may benefit from multiple opportunities to demonstrate skills when inattention interferes, or they have difficulty engaging. It is especially important for clinicians to pair performance on developmental measures with parent-report since assessments completed by clinicians alone may not completely capture a child's full range of skills. Similarly, reporting on skills that are observed during an evaluation, but outside of the developmental test, may also provide a more complete picture of a child's functioning. For example, a clinician might consider observing and noting receptive and expressive language skills that are observed throughout an evaluation (e.g., during informal behavior observations) rather than only what is seen during administration of a given assessment.

Lastly, those with FXS will likely benefit from clinicians moving between developmental domains to administer items. For instance, shifting between cognitive items (e.g., administering a puzzle item) and language items (e.g., identifying pictures in a book), often maintains engagement more effectively than requiring or expecting a child to complete all items in a single domain in consecutive order.

Such flexibility allows the assessor to follow a child's lead, encourage sustained motivation and attention, and improves overall rapport. Note that, depending on the assessment/test, the examiner may not have the flexibility of changing order of items. If the examiner breaks standards, they will likely put this in the narrative of the report.

Limitations

One of the primary limitations of developmental testing is that results represent only a snapshot of a person's full skillset. It is often difficult for clinicians to fully capture the range of skills a person has based on a measure that is typically completed in 45-60 minutes especially if behavioral factors exist such as aggression, tantrums, sensory processing issues, self-harm, hunger, thirst, and/or fatigue). When present, developmental testing will often underestimate true abilities, and thus it is especially important to pair results from developmental testing, as noted above, with caregiver-report.

Assessment from Multiple Perspectives, Sources, and Settings

As with any assessment, collecting information from multiple sources, perspectives, and settings will provide a more reliable and accurate measurement. This may be especially critical for people with FXS who can show highly variable behaviors and abilities depending on mood states, features of the environment (i.e., loud, crowded or novel), timing of medications, or how comfortable and familiar they are with the assessor. It may be important to consider whether testing over one or multiple testing sessions is appropriate.

In all testing areas, collecting observations and/or ratings from more than one caregiver and a rater outside the home, such as a teacher, is encouraged so as to minimize any unintentional bias resulting from having just a single rater. For example, in the cognitive domain, one should try to obtain records of prior testing, and include assessments of diverse areas of function including executive function, verbal comprehension, processing speed, memory, and arithmetic reasoning.

In sum, the administration and interpretation of one assessment tool should never pass for a thorough evaluation. Instead, the combination of thorough record review, family interview and school/vocational placement report, performance on appropriate standardized measures, behavioral observation and educated and thoughtful clinical judgment is necessary for comprehensive assessment.

Preparation and Approach to Testing

General Information

Many preparations, strategies and accommodations can be used to maximize the chances for a successful and valid clinical assessment of a person with FXS. These considerations are described in more detail below.

Unique aspects of FXS are anxiety, hyperarousal and sensory processing difficulties. It is critical to consider these during testing. Many individuals are already anxious prior to the start of an evaluation. Eye-contact, touch, unexpected noise, as well as the fatigue that may come with performing physically and emotionally difficult tasks can all lead to increased hyperarousal. Paying attention to signs such as increased perspiration (especially in the palms), redness in cheeks or ears, rapid breathing, increased gaze avoidance or attempts to avoid the task with conversation (often in higher functioning or more verbal individuals) can help the assessor intervene earlier (i.e., minimize eye contact, use side conversations, increase breaks, offer rewards, etc.) to reduce anxiety and hyperarousal. In a comprehensive clinical assessment, various tasks can be interspersed in the evaluation with intentional breaks that include opportunities to move. The assessor should pace the testing session to maximize performance and maintain ease and engagement of the person being tested. Experienced assessors familiar with the needs of those with FXS are adept at taking proactive breaks to maintain the pace and flow and allow for a productive assessment experience.

Observing the child's response patterns while attempting a task is valuable in assessing how the child solves a problem. This provides valuable diagnostic information when developing school strategies to improve learning.

In general, parents and other caregivers, and those conducting the assessment, should always be sensitive to the child's need for breaks and rest, the need for food and beverage breaks, and a comfortable environment free of distractions and neither too warm nor too cool. Except for the very youngest of children, who may not understand verbal descriptions of a planned visit to a unique setting (if that is the case), children, including older children and adults with intellectual disability, may benefit, and have lessened anxiety if they receive an advance description of what will be happening. However, in some cases, such a description may actually increase anxiety and, therefore, parents will need to make a judgement call about how much and when they should begin to prepare their child for an assessment session. Lastly, the assessor should allow appropriate time for the individual to become comfortable in the testing environment and to develop appropriate rapport, prior to starting the assessment.

Presenting tasks with clear verbal and visual supports that denote beginning and end can be particularly useful when assessing individuals with FXS. For example, provide empty boxes that are crossed off (or filled with a sticker of their choice) after the completion of a task or demonstrate the goal/end of the task (not just the beginning/how to do it or start it). Examples: "when all the coins are in the box" or counting together to ten.

Consideration of the testing environment itself also is imperative, as sensory sensitivities and hyperarousal can impact performance. Reducing the amount of light, closing blinds to eliminate visual distraction, positioning the assessor between the individual and the door, and decreasing auditory interruptions (announcements over a speaker, loud fans or clocks, etc.) will help to encourage the best effort of the person being assessed by promoting focus and sustained attention. Utilizing the expertise of an occupational therapist on the team to train the assessment team on how to support sensory needs and manage anxiety and hyperarousal can be very helpful.

Young children with FXS may be hyperactive and may be better able to respond to test items if they are not confined to a chair; others will benefit from the added structure and confinement of a seated position. These young children often need very many breaks, and all individuals with FXS may do best with testing divided across days to minimize fatigue and frustration.

Many people with FXS respond well to humor or to comments about favorite interests, which can help to reduce anxiety and improve rapport.

Prior to the test

The assessor should obtain information from the caregiver about expected challenges and past experiences with testing and should collect information about items or activities that may be especially motivating to use as rewards during the testing process. It will be important to communicate with the caregiver about what to expect of the testing environment and schedule. Some assessors utilize a visual schedule that can be sent ahead of time to preview what will happen, to reduce anxiety and increase predictability. To minimize anxiety and increase rapport, one can send a friendly photo or a link to photos, or a short video of the assessor and testing environment before the testing day. People with FXS are often remarkable in their ability to recall names and faces and this can be used to ease anxiety, as they will often recognize the assessor immediately upon introduction.

It is important to establish whether the caregiver will be present in the testing room. Some younger children or especially anxious individuals may need this to even enter the testing room, and thus will require the caregiver to be present throughout. Others will do much better without the caregiver present. Finally, a third group seems to do well with the caregiver initially present and then excused. Flexibility from the assessor is crucial, and the assessor may have to use different strategies throughout the process. For example, inclusion of the caregiver and multiple breaks may be successful at the beginning of testing, but the caregiver later may need to leave, and the assessor may need to reduce the numbers of breaks in order to maintain momentum and to not lose attention and motivation.

The assessment team may consider gathering information about interest areas and favorite topics so that they can personalize materials and interactions so that there is a sense of familiarity readily available in establishing rapport.

Test Day

On test day, the person being assessed should be as rested as possible, adequately fed (and prepared with snacks), equipped with any necessary vision correction (glasses, contacts) and/or communication devices, physically healthy, and following their typical medication protocol, to name a few considerations. The amount of time for testing may be either much shorter than usual for a typically developing person (if the person with FXS is unable to progress very far on test items), or it may be much longer than usual (if the person is agitated, needs many breaks, etc.) so it is best to schedule more time than typically expected if possible, and to be flexible regarding breaks and pauses in administration of the assessment.

Accommodations that can be Made to the Testing Process

A visual schedule can be quite helpful whereby the person being assessed can see tasks that must be completed visually depicted in a sequence (using either photos or symbols) and is allowed to cross off completed tasks. A practice or warmup period may be helpful.

Initial testing items should be selected that are easy and they should be organized and already displayed for the individual so that testing can commence immediately to generate success right from the beginning. Keeping a good pace is also important to reduce anxiety caused by waiting on the clinician to get materials organized and ready to proceed. A detailed explanation of the testing process is often unhelpful and may increase anxiety.

Inserting breaks into testing, where access to favorite interests or preferred toys can be granted, may help to support motivation. However, special consideration should be given to whether the individual will be able to successfully transition back to structured activities if they are allowed access to preferred objects or interests.

When utilizing accommodations or "breaks from standardization" during testing, the key concept to keep in mind is to provide adjustments to the testing process if doing so will allow the person with FXS to demonstrate their ability and knowledge without altering the items or skills being measured. For example, it may be necessary to provide more practice items or instructions to ensure understanding of a task, or the assessor may need to repeat an item if the person is distracted. Some standardized tests allow for practice as part of the protocol.)

Use of Rewards to Encourage Participation of the Individual Being Assessed

Additional accommodations may include use of a token economy system (i.e., small rewards throughout testing leading to a bigger prize upon completion), using simplified instructions, allowing a fidget toy or other object, and offering encouraging praise for appropriate effort. When using a token economy system or other reward-based system, it is often important to find out from the caregiver beforehand what has worked with the individual in the past (e.g., sticker vs. goldfish snack) and to consider whether certain rewards may be too distracting and disruptive (e.g., a tablet). When possible, it may be extremely helpful for the individual to identify rewards from a "menu" to ensure cooperation and motivation.

ASSESSMENT DOMAINS

Cognition

Assessment of cognitive function typically includes Intelligence Quotient (IQ) testing and various domains such as executive function (attention, inhibitory control, cognitive flexibility, working memory), processing speed and visual perception, which is the ability to interpret, understand, and define incoming visual information. The assessment process may include activities such as completing puzzles and having individuals answer questions about what they see.

IQ and other cognitive tests can be a very important component of an overall assessment and they provide caregivers, teachers and other providers a sense of developmental level. These measures can be a useful predictor of daily functioning and level of independence, and they can help to form improved treatment, curriculum and transition plans.

It is quite common for IQ scores to decline over time in children with FXS. This is usually not because of a loss in ability – in fact, skills often increase across development, but just not as quickly as a typically developing child. Thus, as the "gap" between FXS development and typical development widens over time, these scores often decrease.

Note that the use of IQ tests for special education eligibility and placements varies from state to state. Many IQ tests are available, and each test has different strengths and weaknesses. IQ tests generate standard IQ, index, or composite scores, which are almost always represented along a normal distribution where 100 is the average score in the general population. It is important to recognize that tests use many different types of tasks to generate these overall IQ scores and some tests have been researched more thoroughly in individuals with FXS than others, including adjustments or accommodations in the administration or scoring that greatly improves their accuracy and can eliminate floor effects (When the person gets the lowest possible score on the tests so their true level of ability cannot be measured.)

Recommended Assessments - Intelligence or Measures of Early Development

- Stanford Binet-5
- Wechsler Scales
    - *Note: Best used for females and higher functioning males.*
- Leiter International Performance Scale-3
- Differential Abilities Scales 2nd edition (DAS-2)
- Kaufman Assessment Battery for Children II

Recommended Assessments - Executive Function

Working Memory:

- Wechsler Digit Forward and Backward
- SB-5 Sentence Memory
- Repeatable Battery for the Assessment of Neuropsychological Status (RBANS)
- Woodcock Johnson Tests of Cognitive Abilities - Memory for Words

Visual Memory:

- Leiter-R Spatial Memory
- SB-5 Block Span
- K-ABC II Atlantis

Assessment of Executive Function Using Caregiver and Teacher Rating Scales:

- Behavior Rating Inventory of Executive Functions (BRIEF): Preschool Version and School-Age Versions
- Conner's Parent Rating Scales
- SNAP-IV

Behavior

Adaptive Behavior / Daily Living Skills

The terms, "adaptive behavior," "daily living skills," "adaptive skills," "functional skills," and "adaptive functioning," are often used interchangeably. They encompass multiple areas of functioning separated into three domains: conceptual skills, social skills, practical skills. Among other skills within each of these domains, communication and socialization, personal self-care skills, and domestic and community living skills are emphasized. Adaptive behaviors include real-life skills such as grooming, getting dressed, following school rules, managing money, cleaning, and making friends. Measuring an individual's adaptive behavior is a critical component as over the last several years, adaptive behavior is increasingly emphasized as an important criterion for defining intellectual disability.

Furthermore, severity of intellectual disability is now characterized by delays in adaptive behavior and not IQ in the Diagnostic and Statistical Manual of Mental Disorders (DSM-5). In addition to aiding in diagnosis, results from adaptive behavior measures may also be used to determine special education eligibility, to plan and implement intervention and rehabilitation services, and to track and monitor progress. Given the reliance on measures of adaptive behavior, reliable and valid standardized assessment is increasingly important.

Recommended Assessments

- Vineland Adaptive Behavior Scales, 3rd Edition (Vinland-3)
- Adaptive Behavior Assessment System, Third Edition (ABAS-3; Harrison & Oakland, 2015)

Maladaptive Behaviors

Maladaptive behaviors include challenging behaviors such as aggression, temper tantrums or self-injury. Maladaptive behaviors are defined as actions or tendencies that do not allow an individual to adjust well to certain situations. They are typically disruptive and can range from mild to severe.

Maladaptive behaviors may be attempts to communicate discomfort (as when asked to make a quick transition or to do an activity they do not want to do), or to reduce anxiety and hyperarousal. Identifying and quantifying these behaviors is a critical component of assessment in FXS as it can aid in tracking response to treatment, as well as in identifying targets for intervention and possible obstacles to learning.

Assessment Strategies

The importance of and reliance upon caregiver-report in maladaptive behavior assessment cannot be understated. As such, an important step in the administration of questionnaires can be to discuss the time period for reporting. (e.g., Discuss what was going on in the household 30 days ago). Providing examples when specific behaviors are not described in the questionnaire or interview can help to improve the accuracy of ratings.

Functional Behavior Assessments (FBA)

These assessments are an in-person observation and data-collection tool that can also be very useful in determining why certain behaviors may be occurring. These assessments can help to determine if certain behaviors are specific to certain environments, which can help to inform the overall diagnostic picture. FBA's can also help to track behavioral change over time, which may help to identify shifts in an individual's behavior that may be suggestive of an underlying concern.

An FBA typically starts with interviews with the child's caregivers (parents and educators), where the specific maladaptive behaviors are described in detail. The interview also identifies antecedents, or things that occur right before the maladaptive behavior and make them more likely to occur, as well as consequences that maintain the behavior and make them more likely to occur. With the maladaptive behaviors defined, direct observations of the individual are conducted in the environments in which the maladaptive behaviors typically occur. These observations help identify the pattern of antecedent-behavior-consequence, which then can be used to determine the function, or purpose, of the child's behavior. This information is then used to identified replacement skills that the child can perform instead of the maladaptive behavior to get their needs met.

Limitations

The reliance upon caregiver questionnaires for assessment of behavior can be a limitation, as various forms of bias and limited recollection of behaviors can affect results. Additionally, the majority of measures have no way of accounting for the level of disruption to the individuals, their family or others. This is especially important when considering treatment intervention and prioritization, as self-harm behavior occurring

only once a day is different than a relatively harmless repetitive behavior like stimming or perseverative questioning. FBA does gather more detailed information than checklists on the frequency, intensity, and duration of maladaptive behaviors, but limitations are that they are time-consuming and require specialized training and experience.

Recommended Assessments

- Aberrant Behavior Checklist-Community, Second Edition (ABC-C-2)
- ABC-Fragile X scoring method (ABC_{FX})

Note that these are just two different ways of scoring the same questionnaire.

Parent/Caregiver Report:

- Behavior Problems Inventory (BPI)
- Restricted and Repetitive Behavior Scale-Revised (RBS-R)
- Vineland Adaptive Behavior Scales-Third Edition (Vinland-3) Maladaptive Domain

Social & Emotional Functioning

The social-emotional phenotype includes prominent symptoms of anxiety (notably social anxiety and specific phobias), irritability or emotional lability, obsessive or perseverative thoughts, and poor coping mechanisms.

Anxiety

The presence of anxiety is common in FXS and has been previously documented at rates higher than other ID populations. The higher rates of anxiety in FXS compared to general ID suggests that the *FMR1* full mutation that causes FXS presents an increased risk for anxiety and has been found to be independent of other clinical factors (i.e., ID, autism diagnosis, gender, age, etc.). As such, a thorough clinical assessment and treatment of anxiety should be included in the FXS standard of care.

Assessment Strategies

Clinicians will often observe that individuals with FXS exhibit heightened anxiety symptoms when initially meeting an individual or in a new situation. As such, it is important to allow time for the individual to become more comfortable in order to assess how their behavior may change with time. Additional probes regarding whether preparation for an event or situation helps or hinders an individual's success, frequency of avoidance and withdrawal, as well as discussion of the sequence of events leading up to aggressive or hyperarousal episodes will help bring clarity to possible anxiety-provoking scenarios.

Note regarding the assessment of anxiety:

Assessments of anxiety in persons with FXS almost always must rely heavily on caregiver report. An exception to this might include a higher functioning male or female who has relatively good insight into their emotions and body sensations, and who can verbalize well. Getting information from multiple sources (mother, father, teacher, other) usually leads to the most comprehensive picture of anxiety symptoms. Though there is no "test" that can be given to measure anxiety in FXS, the assessments below can help bring clarity and insight to an individual's anxiety.

Recommended Assessments (with caregivers)

- Anxiety Disorders Interview Schedule-Parent (ADIS-P)
- Anxiety Depression and Mood Scale (ADAMS)
- Pediatric Anxiety Rating Scale-Revised (PARS-R)

Academic Skills

The assessment of academic skills, also referred to as educational testing, is important for educational and vocational planning. Academic skills pertain specifically to reading, writing, and math skills. Obtaining information about current levels of academic functioning is essential for identifying learning goals, selecting appropriate curriculum materials, informing objectives for instruction, and determining the need for additional intervention and related services (e.g., assistive technology, occupational therapy, environmental modifications, etc.).

Often academic testing is used in an Individualized Education Program (IEP) in school, which provides goals, objectives, accommodations and modifications to a curriculum to meet the unique needs of a student (Refer to the General Educational Guidelines for Students with Fragile X Syndrome in the Treatment Recommendations section of www.fragilex.org for additional information.)

Academic skills are assessed using standardized measures that are norm-referenced and provide similar types of scores as IQ tests. Qualitative measures, such as curriculum-based assessments, that are based on academic content specifically taught in the classroom, may also be used. For children with FXS, it is often important to supplement the results of standardized testing with informal assessment and work samples.

Note: Children with FXS often outperform predictions made about their academic functioning based on their score on an IQ test. Academic functioning can be influenced by environmental factors (e.g., size of classroom, location of seating, amount and type of noise, unusual smells, anything that might affect sensory processing), and presentation

of materials. Additionally, the presence of ASD can impact acquisition of academic skills in FXS.

Assessment Strategies

Strategies to consider include breaking down tasks and changing the presentation of instruction in a step-wise fashion, using visuals (e.g., to assess math concepts), and using additional cues to support how a student approaches a task. These strategies are not considered "standardized," but they can really help facilitate understanding of what the person is able to do under the most optimal circumstances.

Recommended Assessments

- Wechsler Individual Achievement Test-II (WIAT-III)
- Woodcock-Johnson Tests of Achievement-III (WJ-III Tests of Achievement)
- Kaufman Test of Educational Achievement-2nd Ed. (KTEA-II)
- Oral and Written Language Scales, Second Edition (OWLS-II).
- Bracken School Readiness Assessment, Third Edition
- Diagnostic Subtests of the DAS-2

Speech/Language/Communication

Speech refers to the actual production of sounds and words.

Language refers to the content of what we say (i.e. vocabulary/word choice).

Communication is the broadest category of them all and includes not just spoken words, but also nonverbal forms of communication such as crying, reaching, pointing and looking.

Communication abilities are critical to social functioning, as well as to learning about the world in both formal situations, such as school, and informal situations, such as interactions with peers. The inability to communicate can create a cascade in which many aspects of functioning, especially behavioral functioning, are increasingly negatively affected. Assessment of speech, language, and overall communication is important for guiding education, therapy, and even vocational training throughout the life course. There is an array of measurement tools to assess language in individuals with FXS, each with advantages and disadvantages.

Measures and the Use of Standardized Tests

Standardized tests have the advantages of clearly specified procedures. This ensures consistency in administration and allows comparison of the performance of the individual with FXS to typically developing individuals of the same age. Doing so allows for gauging the extent of delay in language for the individual with FXS. A disadvantage, however, is that standardized assessments are often not sensitive enough to measure subtle gains in language skills across time, particularly during adolescence and adulthood, or in response to treatment. Another disadvantage of such tests is that they measure language in situations very different from everyday social interactions, which means that performance on such a test may not always be a good indicator of how an individual with FXS actually uses and understands language or communicates in situations that are personally meaningful, such as school or on the job.

Assessment Strategies

The examiner should select a test based on the skills currently being displayed by the individual rather than strictly by chronological age.

Recommended Assessments

- Preschool Language Scales-Fifth Edition (PLS-5)
- Clinical Evaluation of Language Fundamentals-Preschool 2nd Edition (CELF-P2)
- Clinical Evaluation of Language Fundamentals 5th Edition (CELF-5)
- The Comprehensive Assessment of Spoken Language-2nd Edition (CASL-2)

Parent/caregiver report

- MacArthur Bates Communicative Development Inventories (MB-CDIs) - For earlier stages of language development allow tracking of current and emerging communication skills
- Children's Communication Checklist-version 2 (CCC-2; Bishop, 2003) - For children between the ages of 4 and 16 years who are using phrase speech (e.g., at least three- word utterances)

Natural Communication Sampling

Natural Communication Sampling procedures involve collecting and analyzing audio- or video-recordings of samples of spoken language from an individual with FXS in one or more structured, but naturalistic, interactions with an examiner or adult care provider. It is important that each interaction is structured and scripted so that it is reasonably consistent every time the individual is assessed. When consistency is ensured, natural communication samples can provide excellent measures of an individual's communication skills, including not only spoken language, but also the use of gestures and vocalization in prelinguistic or minimally verbal individuals. Note: Assessment of

natural communication sampling is a general approach to assessment. It can be used to provide a comprehensive analysis of the person's language and help identify targets for language intervention. It can also provide a rough estimate of the extent of delay.

The Occupational Therapy (OT) Practice Framework: Domain and Process (American Occupational Therapy Association, 2014) defines the domain of practice of the profession. Occupational therapy's domain encompasses those areas that parents and research reports tell us create the most difficulty in daily life situations for children, teens, and adults with FXS: play, social participation, sleep, family routines, independent living, and employment. Within this scope, assessments for sensory and motor skills as foundations for all of these areas of daily life function are often included in the OT assessment. OT's have expert knowledge and skills in addressing these areas of occupational performance, including the contextual factors that influence performance, and they use a holistic view of the person and all of his or her interrelated systems and functions. Thus, the profession of occupational therapy, with its broad scope and focus, offers families numerous possibilities for assistance.

Neuromotor Functioning

Neuromotor development and functioning includes many aspects, such as general strength and motor skills, as well as motor speed/response time, dexterity, precision, balance and coordination. Neuromotor skills are integral to the development of many other skills including daily living skills, cognition and language. Note that while the MD or a psychologist may conduct neuromotor testing, an occupational therapist and/or physical therapist is best trained to administer and interpret neuromotor assessments.

Measures

Neuromotor assessments evaluate motor functions through the age at which a skill is expected to be mastered. Many comprehensive developmental tests include the domain of motor skills, especially for infants and toddlers. Even though these tests may be administered "out of age range", they can provide very useful information. For example, an age equivalent score can tell us that the person's motor function is most like a typically developing child of a given age.

Assessment Strategies

There are helpful strategies to consider when directly assessing neuromotor functioning in FXS. As it relates to standardized administration, verbal instructions may need to be adjusted to better match the language abilities of the individual. For example, pointing and saying, "leg up" while demonstrating on the mirrored side (i.e., examiner's right leg for examinee's left leg) instead of "lift your left leg up." More teaching or demonstration

examples may need to be provided. Within the test battery suggested for neuromotor testing, the Movement Assessment Battery for Children (MABC) is unique in that it includes photographs of the items. This type of support increases the non-intrusive guidance that allows for better test administration. Finally, as prompting and guiding is needed, it should be noted that prompting should move from least intrusive (i.e., use of a visual model prior to any other guidance; add gesture or verbal guidance, depending on task) to most intrusive (i.e., physical touch of any kind including hand over hand) to provide the individual with the opportunity to perform the skill in the most independent manner possible. This successive prompt approach can also provide the clinician information as to what type of supports may lead to success in intervention and therapies.

Recommended Assessments

- Movement Assessment Battery for Children-Second Edition (MABC-2)
- Quick Neurological Screening Test-Third Edition (QNST-3)
- Vineland Adaptive Behavior Scales-Third Edition (VABS-3) Motor Skills Domain (parent report)
- Clinical Observations of movement skills (as is common for OT and PT assessments) and may include looking at movement skills of the eyes, body, limbs, mouth and hands is important to complement any standardized testing and to aid in interpreting test results as well as making well rounded recommendations. The testing should results should summarize general motor functioning as well as give information about general gross and fine motor capacities. Also, testing in this area often identifies need for referral to ophthalmology if there are ocular/eye-motor needs identified.

Sensory Processing and Self-Regulation Functioning

Assessment of sensory processing and self-regulation capacities is important for individual's with FXS due to the specific phenotypic impact on these areas.

Measures

- Sensory Processing Measure
- Sensory Profile
- The Body Perception Questionnaire (BPQ)
- Multidimensional Assessment of Interoceptive Awareness (MAIA)

General Occupational Therapy Assessments:

- School Function Assessment (SFA)
- Pediatric Evaluation of Disability Inventory— Computer Adaptive Test (PEDI-CAT)
- Children's Assessment of Participation and Enjoyment/Preference for Activities of Children (CAPE/PAC)
- Transdisciplinary Play Based Assessment

OTHER IMPORTANT CONSIDERATIONS IN THE ASSESSMENT OF INDIVIDUALS WITH FXS

Infant and Toddler Development

Assessment of developmental delays is recommended to occur as soon as differences are noted and/or a diagnosis of FXS is made. When FXS is identified prenatally or early in a child's life, it is often encouraged for these children to be seen by a Fragile X specialist every 3 to 6 months before the age of 2, and at least yearly following their second birthday. Reevaluation of an individual's skills is needed to inform treatment and education plans, and to track progress over time.

Measures

Comprehensive developmental measures assess skills across domains including early cognitive skills, (i.e., problem solving), language skills (receptive and expressive), and motor skills (fine, and gross). While developmental measures do not directly assess social skills and play, as a unique domain, these skills are often captured across items in the cognitive and language domains. For example, participation in social routines (e.g., peekaboo), attending to nursery rhymes, and engaging in back-and-forth play are assessed in the language domain, whereas functional and pretend play skills are assessed in the cognitive domain. In addition, a child's general approach to structured adult-led tasks can also be observed and informally assessed during developmental testing.

Recommended Assessments

- Bayley Scales of Infant Development

Autism

FXS is the most common, known single gene cause of autism spectrum disorder (ASD). While FXS accounts for an estimated 1% to 6% of all ASD cases, many individuals with FXS are co-diagnosed with ASD. Readers should review the 2020 updated version of Autism Spectrum Disorder in Fragile X Syndrome at https://fragilex.org/our-research/treatment-guidelines/ for a more in-depth understanding of the relationship between autism and FXS.

Measures

ASD is defined by the presence of specific social communication and behavioral criteria, and, as such, assessment for ASD, which can be associated with FXS, typically includes a combination of caregiver report and in-person behavioral observation by a specialist knowledgeable about the relationship between ASD and FXS.

Assessment Strategies

For those who are first diagnosed with FXS, pursuing an autism specific assessment often calls for a different approach to evaluation and intervention, primarily due to the presence of relative strengths in social interest and motivation, as well as increased symptoms of hyperarousal and anxiety. Often these increased symptoms of hyperarousal and anxiety may manifest as extremely difficult externalizing behaviors, such as complete withdrawal from social interaction, task refusal, or severe aggressive or self-injurious behaviors. Therefore, additional considerations need to be made when conducting an autism specific assessment.

Recommended Assessments

- Autism Diagnostic Observation Schedule, 2nd Edition (ADOS-2)
- Screening Test for Autism in Two-year-olds (STAT)
- Autism Screening Instrument for Educational Planning, Third Edition (ASIEP-3)
- Autism Diagnostic Interview-Revised (ADI-R)

SUMMARY

The assessment of an individual with FXS must be comprehensive and accommodate the unique aspects of the person. Assessment must utilize tools that are appropriate (feasible, scorable and valid) for use in FXS and commensurate with the individual's developmental level. Professionals are continually working to improve the assessment process and guide the selection of the most appropriate measures for use in FXS and recommendations will be updated accordingly.

Throughout the assessment process, assessors should always remember that individuals with FXS possess numerous strengths such as humor, desire to engage, social interest, visual memory, and sensitivity to others. Whenever possible these strengths should be noted and reflected within the assessment results.

Author Note

This document was excerpted by Robert Miller and Jayne Dixon Weber from a more comprehensive document (see link on page 1) authored by David Hessl, Lisa Cordeiro, Adrienne Villagomez, Elizabeth Coan, Leonard Abbeduto, Anne Hoffman, Angela John Thurman, and Lauren Schmitt. The document represents the current consensus of the members of the Fragile X Clinical & Research Consortium and includes specific edits by FXCRC members Holly Harris, Marcia Braden, Tracy Murnan Stackhouse, Barbara Haas-Givler and Amy Esler.

The Fragile X Clinical & Research Consortium was founded in 2006 and exists to improve the delivery of clinical services to families impacted by any Fragile X-associated Disorder, and to develop a research infrastructure for advancing the development and implementation of new and improved treatments. Please contact the National Fragile X Foundation for more information at (800) 688-8765 or www.fragilex.org.