

Consensus of the Fragile X Clinical & Research Consortium on Clinical Practices

Assessment of Fragile X Syndrome



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The expression of fragile X syndrome (FXS) can range from minimal impact on everyday functioning to significant developmental delays and cognitive deficits, academic problems, social anxiety, depression and autism spectrum disorders. The most effective and available methods for treating FXS are largely provided through special education and various forms of mental health treatments. In order to provide the best educational, therapeutic and vocational programs, careful and thorough psychological assessment is essential. Cognitive strengths and weaknesses, adaptive functioning, communication, academic skills and behavioral issues must all be evaluated on an individual basis in both children and adults, with a multi-disciplinary approach being optimal at all ages. It is very important to integrate information about cognitive, adaptive and behavioral functioning with information obtained from medical status and physical assessments to develop comprehensive interventions. Medical and physical assessments will be discussed in detail under physical problems care consideration.

Cognition

There are a variety of measures for directly assessing cognition in children and adults. For infants and toddlers, the Mullen Scales of Early Learning, the Bayley Scales of Infant Development-3rd Ed or the Merrill-Palmer Revised all provide normative information. Each instrument has pros and cons, with the Mullen and Merrill-Palmer Revised having the advantage of providing cognitive scores based entirely on nonverbal abilities. The Mullen and the Merrill-Palmer Revised can also be used with preschool children, although the Stanford Binet Intelligence Scale-5th Ed (SB-5) and the Differential Abilities Scales-2nd Ed (DAS-2) are often preferred, as they cover a much wider age range from preschool to adults (ages 2-85 for SB-5 and ages 2 ½ -18 for DAS-2). Both provide separate scores for verbal and nonverbal reasoning, and the wide age range of the SB-5 makes it particularly useful with older individuals who function within the lower levels of ability. The Leiter International Performance Scale – Revised is an entirely nonverbal measure of cognition for individuals from preschool to adults, making no demands on either receptive or expressive language. For more able individuals, there are different versions of the Wechsler scales, with separate versions for young children 2 ½ - 7 years (Wechsler Preschool and Primary Scales of Intelligence-3rd Ed), children through early adolescence 6-16 (Wechsler Intelligence Scale for Children-4th Ed) and adults (Wechsler Adult Intelligence Scale-4th Ed), as well as the Kaufman Assessment Battery for Children-2nd Ed for ages 3-18. The Wechsler scales provide separate scores for verbal and nonverbal reasoning while the Kaufman yields a nonverbal composite and is designed to minimize verbal instructions and responses.

Adaptive Functioning

In addition to cognition, measures of adaptive function provide needed information about an individual's everyday skill set. Adaptive measures rely on input from caregivers who are familiar with the individual—usually parents and teachers. Information is typically obtained via a structured interview, and while checklist forms exist, they are often difficult for respondents to complete without assistance. The Vineland Adaptive Behavior Scales-2nd Ed., the Adaptive Behavior Assessment System-2nd Ed. and the Scales of Independent Behavior-Revised each provide a score to capture global functioning for individuals birth to age 90, but also provide scores across several domains, including communication, socialization, daily living, motor skills and maladaptive behaviors to allow identification of strengths and weaknesses.

Communication

Assessment of communication skills should include testing of receptive language (listening), expressive (speaking) vocabulary, syntax and fluency, and pragmatic skills. At all ages, optimal assessments include a combination of formal standardized tests, checklists, and structured and unstructured observations, requiring input from both a speech and language pathologist (SLP) and a psychologist. For example, the Preschool Language Scale-4th Ed., Test for Auditory Comprehension of Language-3rd Ed., the Clinical Evaluation of Language Fundamentals-4th Ed. and the Comprehensive Assessment of Spoken Language are typically administered by an SLP. However, other measures such as the Oral and Written Language Scales (OWLS) and picture vocabulary tests, can be administered by either an SLP or a psychologist. The OWLS includes separate measures for oral language (ages 3-21) as well as a very structured measure of written language (ages 5-21). Structured interviews and checklists assessing social communication skills and formal measures of pragmatic skills are typically administered by psychologists. Measures of social communication are especially important given the incidence of autism spectrum disorders, poor eye contact, and repetitive speech seen in FXS. The Autism Diagnostic Interview-Revised (ADI-R) and the Autism Diagnostic Observation Schedule (ADOS) provide detailed information about communication in addition to determining whether an individual demonstrates characteristics consistent with ASD. The ADOS has four levels ranging from young nonverbal children to verbally fluent adults. For very young children ages 2-3, the Screening Test for Autism in Toddlers (STAT) is a play-based measure for assessing social communication skills. Checklists to assess social communication, severity of autistic behavior, and to screen for ASD, include the M-CHAT (modified checklist for autism in toddlers), the Parent Checklist from the CSBS for young children, and the Social Responsiveness Scale (SRS) and the Social Communication Questionnaire (SCQ) for children and adolescents.

Maladaptive Behavior

In addition to assessing behaviors associated with ASD (poor eye contact, deficits in peer relationship, repetitive behavior, restricted interests), it is important to examine other dimensions of problematic behavior that are even more common in FXS. Aberrant or maladaptive behaviors are typically classified as internalizing (e.g., anxiety) and externalizing (e.g., hyperactivity). They are assessed with checklists completed by parents and other caregivers such as teachers. The Child Behavior Checklist and Caregiver-Teacher Report Form (Achenbach) cover the ages 1 ½ through adults, while the Behavior Assessment System for Children-2nd Ed. (BASC-2) covers ages 2-21. These broad-based measures yield global and domain scores. The Aberrant Behavior Checklist-Community, the most widely used and probably the most useful for the range of cognitive function in FXS, assesses problem behaviors in developmentally challenged individuals ages 6-54 across multiple settings (home, school, clinic, residential facility), including irritability, lethargy, stereotypy, hyperactivity, and inappropriate speech. The checklist can be completed by parents, special educators, psychologists, direct caregivers, nurses and others with knowledge of the individual being assessed.

Academic Skills

The assessment of academic skills is important for educational and vocational planning. Obtaining information about current levels of performance (e.g., readiness vs. early 2nd grade) is essential for selecting appropriate curriculum materials, identifying goals and objectives for instruction and

determining the need for related services (e.g., assistive technology, occupational therapy, environmental modifications, etc.). When selecting measures it is important to capitalize on the evidence that children with FXS learn and perform better when material is presented in a holistic rather than a sequential manner, and with a structured rather than an open-ended format. For example, the Woodcock Reading Mastery Tests-Revised, Normative Update provides picture cues and uses cloze (fill in the blank) procedures for assessing reading comprehension. Multiple choice formats can be useful with individuals with skills beyond the emergent level (e.g., Gray Silent or Oral Reading Tests). To assess mathematics, the Kaufman Test of Educational Achievement-2nd Ed. (KTEA-II) includes picture supported tasks, many requiring only a pointing response (Math Concepts and Applications). The Oral and Written Language Scales (OWLS) and the KTEA-II assess emergent and more advanced writing skills using familiar tasks (e.g., writing name, copying words, labeling pictures, etc.) in a highly structured manner. In some cases, it is important to supplement the results of standardized testing with informal assessment and work samples.

Author Note

This guideline was authored by Monica Dowling, PHD, and Deborah Barbouth, MD, and was reviewed and edited by consortium members both within and external to its Clinical Practices Committee. It has been approved by and represents the current consensus of the members of the Fragile X Clinical & Research Consortium.

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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.