

Making Sense of Multiple Diagnoses

The bad news is that there is a lot of confusion among parents and even professionals about the meaning of Fragile X in relation to other types of diagnoses that a child receives.

For example, a 6-year-old girl is diagnosed by a developmental pediatrician as having autism and intellectual disability. At age 8, a blood test shows that she has Fragile X syndrome. Now what? Does she still have those other disorders? Does she now have Fragile X, too, in addition to intellectual disability and autism?

Similarly, a 10-year-old boy known to have FXS is diagnosed by a child psychiatrist as having ADHD. Does this mean he has now developed a separate, unrelated condition? Understandably, many parents are confused about the “alphabet soup” of diagnoses used to describe their children.

To address these questions, it helps to review the differences between the developmental and genetic diagnostic systems that lie at the heart of this confusion.

Symptoms vs. Causes

First, it’s important to understand the difference between clinical developmental diagnoses, such as intellectual disability and ADHD, and [genetic diagnoses](#), such as [Fragile X syndrome](#).

Put simply, developmental diagnoses include disorders with specific patterns of learning and behavior that occur during childhood. These are also known as clinical disorders, meaning that they are diagnosed by licensed professionals based on a child’s developmental history and observed symptoms.

The clinical developmental disorders associated with FXS are described in the [Diagnostic and Statistical Manual of Mental Disorders](#) (known as DSM-5 because it’s the fifth edition of the book), which is published by the American Psychiatric Association. There are hundreds of different developmental and psychiatric

conditions in the DSM-5, including well-known ones such as ADHD, autism, and intellectual disability.

The DSM-5 outlines the behavioral and other criteria that must be met to diagnose a particular developmental or psychiatric disorder. For example, [to meet criteria for a diagnosis of ADHD](#), a child must display certain symptoms of hyperactivity, inattention, and impulsiveness. Several of these symptoms must be present by age 12, occur in at least two different settings, are not better explained by another disorder, and reduce the quality of social, school, or work functioning. If a child displays behaviors that meet these and other DSM-5 criteria for ADHD, then they may be diagnosed with that disorder.

It's important to note that clinical developmental and psychiatric diagnoses are not made using blood tests or brain imaging studies, such as MRI, and they generally reveal nothing about the underlying cause of the symptoms. Clinical diagnoses are primarily based on whether or not a person meets the symptom criteria defined in the DSM-5 book, regardless of the cause.

By contrast, Fragile X syndrome is considered an underlying cause (also referred to as etiology) of developmental symptoms. The diagnosis of FXS is based on a highly accurate laboratory test that can detect expansions of the Fragile X (*FMR1*) gene. Large expansions of the gene, known as full mutations, cause *FMR1* to shut down, which significantly affects brain function.

By itself, the diagnosis of an *FMR1* full mutation does not reveal anything about specific developmental and behavioral symptoms an affected child has or will develop in the future. While *FMR1* full mutations cause significant problems with brain function, the symptoms can vary from one person to the next and may fit into one or more different clinical diagnoses.

In fact, virtually all children and adults diagnosed with FXS have symptoms that meet the criteria for at least one, and usually more, clinical developmental disorder. Intellectual disability, autism, and ADHD are among the most common clinical diagnoses made in children with FXS.

Does this mean that individuals with FXS have several different unrelated disorders? Not at all. They almost always have one underlying cause disorder — Fragile X syndrome — that results in patterns of behavior and learning that meet criteria for multiple symptom disorders, including intellectual disability and autism.

Therefore, the 10-year-old boy mentioned above who is subsequently diagnosed with ADHD most likely does not have two unrelated conditions: his clinical diagnosis of ADHD is in all likelihood a symptom of his FXS.

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So, What Does My Child Have?

Once we recognize the difference between symptom and cause diagnoses, it becomes clear that a child or adult may have multiple symptom diagnoses plus a cause diagnosis all at the same time.

Trying to compare these different types of diagnoses is like comparing apples and oranges: they serve different purposes. A Fragile X syndrome diagnosis can answer important questions about the cause of a disability, its genetic implications, associated physical and behavioral symptoms, and potential treatments.

Knowing a Fragile X syndrome diagnosis also allows families to participate in research, including [clinical trials](#), that may one day result in important treatment breakthroughs. Above all, a genetic diagnosis provides the fundamental answer to a parent's question: What does my child have?

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The Importance of Clinical Developmental Diagnoses

Clinical developmental diagnoses are important for other reasons, not the least of which is eligibility for school and therapy services. There are no laws that specifically guarantee an appropriate special education for children with FXS. However, in the U.S. and other countries, laws do exist that give that right to children with certain clinical developmental disorders, such as intellectual disability and autism.

Equally important, clinical diagnoses can guide decisions about psychiatric [medications](#) used for the treatment of behavioral symptoms. For example, stimulant medications, such as the drug Adderall, are not specifically prescribed to treat FXS but are often used successfully to treat attention disorders in children with FXS.

The same holds true for non-medical therapies. A child whose autism is due to FXS may still benefit from educational and behavioral interventions developed for children whose autism is not related to FXS.

The bottom line? Understanding the difference between symptoms and causes allows individuals with FXS and their families to access the best of both diagnostic worlds.

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About the Author

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