Fragile X Syndrome
Patient Clinical Journey

NATIONAL FRAGILE X FOUNDATION | EVERYLIFE FOUNDATION FOR RARE DISEASES

DELAYED DIAGNOSIS

Items in red may be prevented with a Timely Diagnosis.

PRE-DIAGNOSIS (ages 0-5 for boys, ages 0-7 for girls)

Typical Accompanying Diagnoses
- Chronic otitis media/tube placement
- Eye muscle weakness (strabismus) – *May require surgery*
- Gross motor delay/gait issues/lack of coordination
- Loose connective tissues
- Sensory processing issues
- Speech and language disorders/delays
- *The following diagnoses could lead a family to pause or end the diagnostic odyssey due to several factors:*
  - ADHD
  - Autism
  - Global Developmental Delay
- *The following usually present later in the pre-diagnosis stage*
  - Anxiety
  - Epilepsy, partial epilepsy, convulsions
  - Self-Injury/aggressive behavior

Specialists
- Dentist
- Developmental Behavioral Pediatrician
- Ear Nose, & Throat Specialist
- Geneticist
- Neurologist
- Ophthalmologist
- Pediatrician
- Physical Therapist (PT)/Occupational Therapist (OT)/Speech Pathologist (SP)
- Social Worker

Treatments
- *Attention deficits, mood, and anxiety disorders may require evaluation and behavioral/pharmacological interventions. Some examples of pharmacological interventions include SSRI's, stimulants, and alpha-agonists.*
- *Less common interventions may include anticonvulsants and antipsychotics.*
- Early Intervention
- Special education

Testing & Procedures
- Microarray
- MRI
- Neuropsychological testing for cognitive deficits
- Autism Testing – *Generally occurs later in the pre-diagnosis stage*
- Testing for FMR1 mutation – *Leads to diagnosis*

TIME OF DIAGNOSIS (age 5 for boys, age 7 for girls)

Typical Accompanying Diagnoses
- *All previously listed diagnoses may remain as an accompanying diagnosis to Fragile X*

Specialists
- Dentist
- Developmental Behavioral Pediatrician
- Ear Nose, & Throat Specialist
- Geneticist
- Neurologist
- Ophthalmologist
- Pediatrician
- Physical Therapist (PT)/Occupational Therapist (OT)/Speech Pathologist (SP)
- Social Worker

Treatments
- *Attention deficits, mood, and anxiety disorders may require evaluation and behavioral/pharmacological interventions. Some examples of pharmacological interventions include SSRI's, stimulants, and alpha-agonists.*
- *Less common interventions may include anticonvulsants and antipsychotics.*
- Early Intervention
- Special education

While the list of typical additional accompanying diagnoses is representative, families may experience additional challenges/ diagnoses related to GI and sleep.

POST DIAGNOSIS (up to age 15)

Typical Accompanying Diagnoses
- *All previously listed diagnoses may remain as an accompanying diagnosis to Fragile X*

Specialists
- Behavioral and Therapeutic Services
- Child Neurologist
- Child Psychiatrist
- Counseling Support Services
- Dentist
- Developmental Behavioral Pediatrician
- Ophthalmologist
- Pediatrician
- PT/OT/SP
- Social Worker

Treatments
- *Attention deficits, mood, and anxiety disorders may require evaluation and behavioral/pharmacological interventions. Some examples of pharmacological interventions include SSRI's, stimulants, and alpha-agonists.*
- *Less common interventions may include anticonvulsants and antipsychotics.*
- Early Intervention
- Special education

TIMELY DIAGNOSIS

PRE-DIAGNOSIS

Typical Accompanying Diagnoses
- Chronic otitis media/tube placement
- Eye muscle weakness (strabismus) – *May require surgery*
- Gross motor delay/gait issues/lack of coordination
- Loose Connective Tissues; Sensory Processing Issues
- Speech and language disorders/delays
- *The following diagnoses could lead a family to pause or end the diagnostic odyssey due to several factors:*
  - ADHD; Autism
  - Global Developmental Delay
- *The following usually present later in the pre-diagnosis stage*
  - Anxiety
  - Epilepsy, partial epilepsy, convulsions
  - Self-Injury/aggressive behavior

Specialists
- Child Neurologist; Dentist
- Developmental Behavioral Pediatrician
- Ear Nose, & Throat Specialist
- Ophthalmologist; Pediatrician
- PT/OT/SP; Social Worker

Testing & Procedures
- Microarray
- Autism Testing – *Generally occurs later in the pre-diagnosis stage*
- Testing for FMR1 mutation – *Leads to diagnosis*

TIME OF DIAGNOSIS (by age 2)

Typical Accompanying Diagnoses
- *All previously listed diagnoses remain as an accompanying diagnosis to Fragile X*

Specialists
- Behavioral and Therapeutic Services
- Child Neurologist; Dentist
- Developmental Behavioral Pediatrician
- Ear Nose, & Throat Specialist
- Geneticist
- Ophthalmologist; Pediatrician
- PT/OT/SP; Social Worker

Treatments
- Early Intervention; Special education

While the number of specialists and providers may not shift with earlier diagnosis, access to appropriate care is greatly expedited with earlier diagnosis.

Earlier diagnosis also supports the parent who is a Fragile X premutation carrier with both receiving timely care for themselves and evaluating considerations for future family planning.

POST DIAGNOSIS

Typical Accompanying Diagnoses
- *All previously listed diagnoses remain as an accompanying diagnosis to Fragile X*

Specialists
- Behavioral and Therapeutic Services
- Child Neurologist
- Child Psychiatrist
- Counseling Support Services
- Dentist
- Developmental Pediatrician
- Opthalmologist
- Pediatrician
- PT/OT/SP
- Social Worker

Treatments
- *Attention deficits, mood, and anxiety disorders may require evaluation and behavioral/pharmacological interventions. Some examples of pharmacological interventions include SSRI's, stimulants, and alpha-agonists.*
- *Less common interventions may include anticonvulsants and antipsychotics.*
- Early Intervention
- Special education

**This guide is not intended as a clinical decision-making tool. Please consult your specialists for any medical decisions.*