

CYFIP2 EIEE/DEE REFERRAL CHECKLIST

This is a list of commonly required medical services. Not all children will need each service listed.

- **Pediatric Neurologist:** Essential for managing epilepsy and developmental concerns.
- **Epileptologist:** A Neurologist specializing in complex epilepsy and refractory seizures.
- **Geneticist/Genetics Counselor:** Helps families understand the genetic basis of CYFIP2 EIEE, offers guidance on inheritance patterns, prognosis and testing for family members,
- **Developmental Pediatrician:** Monitors and manages developmental delays and disabilities, coordinates therapies and educational interventions.
- **Nutritional/Feeding Specialist:** Evaluates for potential dietary therapies such as the Keto diet for seizure control, manages feeding issues such as oral motor delays, evaluates need for G-Tube placement.
- **Pediatric Hematologist:** Some children with CYFIP2 EIEE/DEE experience severe neutropenia (low neutrophil count) requiring routine monitoring and/or additional medications.
- **Occupational Therapist:** Focuses on fine motor skills, daily living activities and may address sensory integration issues.
- **Physical Therapist:** Focuses on gross motor development, muscle tone and mobility, can provide adaptive equipment if needed.
- **Speech-Language Pathologist (SLP):** Addresses communication challenges, including speech delays and feeding difficulties, implements communication strategies if necessary.
- **Neuro-ophthalmologist:** Diagnoses and manages visual issues related to the nervous system, some children with CYFIP2 EIEE/DEE experience visual impairments such as CVI.

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- **Vision Therapist:** Provides customized exercises and activities to improve visual skills and how the brain processes visual information.
- **Complex Care Coordinator:** Assists in organizing appointments and managing communication between providers.
- **Early Intervention Specialist:** Birth to age 3, provides developmental evaluations and in-home therapies.
- **Education Specialist/IEP Advocate:** Assists with individualized education plans (IEPs) to meet the child's needs in school.
- **Respite Care Provider:** Provides temporary care to give families a break from the demands of at-home medical needs, some respite care programs offer skilled nursing.

When a child is first diagnosed with CYFIP2 EIEE/DEE, families will need to engage with a multidisciplinary team of healthcare providers and specialists to address the complex needs associated with this condition. This list is meant to serve as a guide for families. Not all children will need every service listed, and some children may require additional services. Each child will have their own individual needs.

For more information, please visit:
www.cyfip2network.org

