Project FIND-OUT

What Is Project FIND-OUT?

Project FIND-OUT is a research study that aims to reduce the time to diagnosis in rare genetic neurodevelopmental disorders (RGNDs). The objectives are to understand the impact of early whole genome sequencing on patients, caregivers, and healthcare providers. The goal is to influence the development of national guidelines and payor coverage policies to make it easier for healthcare providers and patients to access whole genome sequencing.

What Services Does Project FIND-OUT Provide?

Your baby will receive a free neurological assessment and whole genome sequencing, and your family will receive genetic counseling. With your consent, your doctor will receive the results from whole genome sequencing and recommendations on next steps, including potential referrals and additional testing. All services are free and available via phone and online - you will not need to travel.

Why Conduct Genetic Testing So Early?

For many rare genetic neurodevelopmental disorders (RGND), less than 50% of patients have a confirmed genetic diagnosis and the average time between parents first identifying a concern to a genetic diagnosis is around 30 months. A long diagnostic journey is difficult for families and may prevent patients from receiving the maximum benefit of gene therapy and other disease-modifying therapies. Even in RGND without therapies (approved or in clinical trials), a genetic diagnosis can impact clinical management.

What Is Required to Participate?

To participate, caregivers will:

- Participate in a one-hour virtual assessment (online via FaceTime or other online platform) which will include reviewing and completing the consent form
- Receive and return the test kit, which will require the caregiver to collect a buccal swab
- Participate in a 30-minute genetic counseling session once the results are available
- Provide feedback on their experience with the project 12 weeks after their genetic counseling session (online plus an optional telephone interview)

What Are Potential Risks of Participating?

The project team will review the risks and benefits of the project with parents/caregivers during the enrollment process. Some of the risks include:

- Some genetic testing may return a “non-diagnostic” result which may require the family to collect another sample or collect a different type of sample in order to receive the results.
- The child may or may not get a diagnosis from WGS. Not every child who meets the Project FIND-OUT criteria will have a genetic cause and not every genetic cause will be identified in testing.
- If the child receives a genetic diagnosis, there may or may not be a treatment available for the genetic disease.

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Which Patients Are Eligible?

Infants 3–12 months of age who meet at least 2 of the criteria below are eligible for Project FIND-OUT.

FIND-OUT Eligibility Criteria

F **Feeding issues**
Inability to suck, reflux, constipation, dysphagia

I **Issues with movement**
Ataxia, Dystonia, including torticollis, Choreiform movements, Tremor, Myoclonus, Stereotypy, Tics, Tremulousness

N **NICU admission**
Any NICU admission for any reason of any duration

D **Developmental delay**
Missed milestones in 2 or more domains including communication or language, gross motor, fine motor, problem solving, personal-social

O **Other** ***(congenital malformations, atypical growth or specialist referral)***
Structural anomalies including dysmorphic craniofacial features or congenital malformations including congenital heart defects • Atypical growth in head circumference, weight, or length • Referral to a specialist (physician, PT/OT/ST)

U **Unprovoked seizures**
One or more unprovoked seizures

T **Tone**
Hypotonia including “floppy baby” or excessive head lag, hypertonia, abnormal deep tendon reflexes or clonus

How Can My Patient Learn More And Enroll?

If you have a family who is interested in the project, the family can go to **www.projectfindout.com** or contact **clinicalteam@projectfindout.com**.