



Benefits of a Genetic Evaluation and Diagnosis for the CHILD

- Early diagnosis and recognition may impact the long-term outcome for children with many conditions.
- Find appropriate medical and non-medical therapies
- Screen for associated complications or disabilities before symptoms are seen
- Avoid unnecessary testing and evaluation
- Help with educational planning (immediate and long term)
- Start physical therapy, occupational therapy, and/or speech therapy, as soon as possible if needed.

Benefits of a Genetic Evaluation and Diagnosis for the PARENTS

- Provide information about the cause and treatment
- Offer support, guidance and an outline of what to expect
- Aids in reducing the sense of helplessness when the diagnosis/prognosis is uncertain
- Enables referrals to appropriate medical and social service agencies, support groups
- Learn about opportunities for education, advocacy and research
- Provides accurate reproductive options, carrier testing, prenatal /preimplantation genetic diagnosis
- Help with decisions about long-term care planning (insurance, education, contraception, adult living)

Benefits of a Genetic Evaluation and Diagnosis for ADULTS

- Children and teens with metabolic conditions transitioning to adult care: care and support can continue without interruption
- An appropriate diagnosis of a genetic disease of adult onset can help guide optimal management and care for that patient
- Medical management can be guided by genetic testing even in pre-symptomatic or asymptomatic individuals. This can be significant and potentially life-saving
Ex. hereditary cancer syndromes
- Genetic tests can indicate if a person will benefit or not from certain medications, the dose that will benefit that patient, and if the person is at risk for an harmful reaction.
Ex. Abacavir (medication for HIV) and carbamazepine (medication for seizures and neuropathic pain)
- For adults with a family history of a genetic condition, carrier testing, accurate reproductive options and prenatal /preimplantation genetic diagnosis can be offered.