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 a 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.