Natural History Study for DNA Repair Disorders

Status: Recruiting

Eligibility Criteria

Sex: Male or Female
Age Group: Not specified
This study is NOT accepting healthy volunteers

Inclusion Criteria:

- at least 6 months old - diagnosis of Cockayne syndrome (CS), xeroderma pigmentosum (XP), or trichothiodystrophy (TTD), based on genetic testing and/or key clinical characteristics

•have one or more of the neurodevelopmental or neurological complications such as gross motor delay, language delay, altered muscle tone (study staff will review) - family member of an individual with the above condition

Exclusion Criteria:

- prior history of systemic gene or cell-based therapy - participation in a clinical trial for treatment

Conditions & Interventions

Interventions:
Other: ECAB Assessment, Other: Gait Assessment, Other: Interval History, Other: Physical Examination, Other: Specimen Sample Collection
Conditions:
Rare Diseases
Keywords:
Cockayne Syndrome, DNA Repair Disorder, Trichothiodystrophy, Xeroderma Pigmentosum

More Information

Description: This research is being done to help us better understand the different DNA repair disorders. We will collect data and samples that we will use to develop new therapies and medicine to help treat the disease. We expect that participants will be in this research study for 3 years. Visits will occur every six months and alternate between in-person and remote. Remote visits should be expected to last 1-2 hours, and in-person visits should be expected to last 3-4 hours.
Study Contact: Erin Aguero - aguer008@umn.edu
Principal Investigator: Peter Kang
IRB Number: STUDY00015911