Natural History Study for DNA Repair Disorders

Status: Recruiting

Eligibility Criteria

Age: Not specified

This study is NOT accepting healthy

Healthy Volunteers: 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

Conditions:
Rare Diseases
Keywords:
Cockayne Syndrome, DNA Repair Disorder, Trichothiodystrophy, Xeroderma Pigmentosum

More Information

Description: A single-center, single-arm, non-interventional natural history study to evaluate the longitudinal clinical course, functional outcome measures, and candidate biomarkers for individuals with DNA repair disorders, including Cockayne syndrome (CS), xeroderma pigmentosum (XP), and trichothiodystrophy (TTD).
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