

Quercetin Dyskeratosis Congenita (DC)/Telomere Biology Disorders (TBD)

Status: RECRUITING

Eligibility Criteria

Sex: ALL

Age: 2 years and over

Healthy Volunteers: This study is NOT accepting healthy volunteers

Inclusion Criteria:

1. Diagnosis of DC/TBD deficiency as defined by at least one of the following:
 - Age adjusted mean-telomere length of <1 percentile in all tested peripheral blood cells such as granulocytes, lymphocytes, B-cells, naïve T-cells, memory T-cells, and NK cells
 - A pathogenic or likely pathogenic mutation in DKC1, TERC, TERT, NOP10, NHP2, TINF2, CTC1, PARN, RTEL1, ACD, NAF1, ZCCHC8, or WRAP53
1. Patients $\geq$ 2.0 years of age*
- The first three enrolled patients must be $\geq$ 10.0 years of age
1. Able to take medication orally

Exclusion Criteria:

1. Renal failure requiring dialysis
2. Total bilirubin >3 mg/dl and/or SGPT >300 at time of enrollment, unless elevation thought to be related to DC/TBD
3. Patients who have received quercetin or any over-the-counter antioxidant supplementation within last 1 month
4. Patients currently taking androgen therapy
5. Patients receiving digoxin therapy, who are unable to discontinue treatment due to medical reasons
6. Patients receiving fluoroquinolone therapy, who are unable to discontinue treatment due to medical reasons
7. Patients who are pregnant or breastfeeding or are at risk of pregnancy and are unable to use acceptable methods of birth control during the length of the study
8. Patients with morphologic or cytogenetic evidence of myelodysplasia or leukemia.
9. Patients needing to start or actively receiving radiation therapy, chemotherapy or immunotherapy for treatment of SCC or other cancers.
10. Patients with unstable disease status or other medical issues requiring hospitalization or rapid escalation of medical care
11. Participating in another therapeutic study for DC/TBD
12. Patients who are in the early post-stem cell transplant period (i.e. first 6 months post-transplant)

Conditions & Interventions

Interventions:

DRUG: Quercetin

Conditions:

Dyskeratosis Congenita, Telomere Disease

More Information

Description:

The purpose of this study is to see if a vitamin-like substance called quercetin is safe for people who have a rare condition called Dyskeratosis congenita (DC) or telomere biology disorders (TBD).

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Principal Investigator:

Principal Investigator ID:

Phase: PHASE1

IRB Number:

System ID: NCT07628972

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