A Natural History Study Seeks to Understand the Clinical, Genomic, Pharmacological, Laboratory, and Dietary Determinates of Pyrimidine and Purine Metabolism Disorders

NCT06092346

Status	RECRUITING
Sponsor	National Human Genome Research Institute (NHGRI)
Enrollment	999 participants

Key Eligibility Criteria

Inclusion (24)

- There are three populations that will be included in this study: subjects with known DPPM, family members of study subjects, and healthy controls.
- In order to be eligible to participate in this study as a subject with a known DPPM an individual must meet all following criteria:
- At least one month of age;
- A medical history that, based on the preponderance of clinical, laboratory, biochemical, and/or genomic evidence is consistent with DPPMs;
- Clinical findings that can be used to suspect disorders of purine and pyrimidine metabolism will include, but not be limited to the presence of congenital malformations, neurological, behavioral, immunological, rheumatological, hematological, renal involvement; gout; and recurrent rhabdomyolysis in one or more family members.

... and 19 more (see full listing online)

Exclusion (4)

- Unrelated volunteers who are unaffected with DPPM but have intellectual disability due to other causes, such that they cannot provide informed consent without a guardian/LAR, will not be enrolled in this study. Affected individuals and family member(s) of individuals with DPPM can participate in the study when appropriate informed consent is obtained (with aide of parents/guardian/LAR/bioethics review when necessary).
- Intercurrent or chronic conditions which in the opinion of the investigators, can then interfere with the interpretation of research studies (e.g. ongoing cancer treatment resulting in bone marrow suppression in a patient with DPPM also presenting with bone marrow suppression).
- Pregnant participants as unaffected family members or as unrelated healthy volunteers are not able to join the protocol during the pregnancy.
- Individuals without a routine clinical care team outside of the NIH cannot enroll in this study. We will ask the participants for the name of clinical care team prior to enrollment.

Locations (1 total)

National Institutes of Health Clinical Center, Bethesda, Maryland, United States