

Study of Proteus Syndrome and Related Congenital Disorders

NCT00001403

Status	RECRUITING
Sponsor	National Human Genome Research Institute (NHGRI)
Enrollment	1,500 participants

Key Eligibility Criteria

Inclusion (7)

- All patients who meet clinical diagnostic criteria for PS, or who have demonstrated AKT1 p.Glu17Lys variants are considered eligible for this protocol. As well, we will generally offer an in-person evaluation at the NIHCC to patients with PS whenever possible.
- As these disorders are usually apparent at or soon after birth, and appear to evolve at least into the third decade of life, early assessment and long-term follow-up are necessary. We have already learned that PS has a high pediatric mortality rate. PS and other overgrowth disorders are progressive and for some individuals, may warrant more frequent observation during youth and adolescence. Therefore, it would not be practicable or ethical to exclude children from enrollment.
- Patients with overgrowth that is not definitively PS (i.e., who do not appear to meet clinical diagnostic criteria) may also be eligible to participate in this study. Decisions to invite patients in this group to the NIHCC for an in-person evaluation are made on a case-by-case basis where the patient's phenotype, health, proximity to the NIH, and fit with our current research aims will all be taken into account. In general, we will consider subjects who have one or more of the manifestations from the PS clinical criteria as eligible.
- Enrollment of adults with impaired decision-making capacity is scientifically justified because PS is an ultra-rare disorder where 10-15% of patients have significant cognitive impairment and gaining a better understanding of this aspect of the phenotype (as well as the other concerns adult patients may present with) is critical to advancing our knowledge of this disorder. Progression of overgrowth, particularly the fibroadipose overgrowth in CLOVES syndrome, is a significant issue in many adults with this condition and understanding the trajectory of overgrowth throughout the lifespan is an important goal of this study.
- This protocol enrolls participants of all ages which includes women of child-bearing age. We recognize that women may become pregnant during the course of this study. While we have not documented a case of a female with Proteus syndrome becoming pregnant it is important to gather clinical data if such a case occurs in order to better understand the natural history of Proteus syndrome and related disorders.

... and 2 more (see full listing online)

Exclusion (3)

- Patients with cancer but who do not have overgrowth or other non-tumor manifestations of PS or non-PS overgrowth, whose tumors may harbor AKT1, PIK3CA, or other variants, are not eligible for this study. In general, patients who clearly meet diagnostic criteria for a well-characterized overgrowth syndrome that is NOT PS are not eligible for this study. Bannayan-Riley-Ruvalcaba syndrome and PHACES syndrome are examples of such entities. We will not enroll prisoners, healthy volunteers, or lab personnel. Some persons with PS and other overgrowth conditions are intellectually disabled (ID) or developmentally delayed (probably ~10%). The consent issues are no different for children with ID than developmentally appropriate children except that assent will be judged by developmental level instead of age. Patients who are adults and decisionally-impaired are eligible only if they have a legal guardian who has authority to sign a consent form on their behalf. Patients who are medically fragile or unable to tolerate travel to the NIHCC will not routinely be eligible for participation.
- We will request permission to retain some information about prospective participants who may not be immediately enrolled. As these participants will not immediately be signing a consent form and joining the study, we propose to NOT count these participants in our Inclusion Enrollment Reports until they have formally enrolled in the study (that is, they have signed consent forms).
- We will not enroll neonates (newborns less than one month old).

Locations (1 total)

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

<https://clinicaltrials.gov/study/NCT00001403>

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