SAMD9/9L Mutational Registry

Current knowledge about SAMD9 and SAMD9L (SAMD9/9L) syndromes is limited and the genetics not well understood. Many of the mutations are not being collected and are unavailable to the public, therefore missing out on valuable data and future education. The purpose of this registry is to learn about new genetic variants and serve as a knowledge base for the medical and research community. Since most new patients have novel mutations, knowledge about other patients across the world is essential. We also wish to expand the phenotypic spectrum of disease that might help to better understand the disease and prevent future complications. The information collected in this registry could also benefit patients through optimal genetic counseling and risk-adapted management.
De-identified data will be submitted from the managing clinician or geneticist. This study falls under 2018 HHS common rule 45 CFR 46.104 (4) exemption: secondary research for which consent is not required. The data will be updated on a regular basis, and once enough data is collected, we anticipate publishing the results as a research manuscript on genetics, phenotypes, and genotype-phenotype associations in SAMD9/9L patients. Submitting investigators will be acknowledged. We will inform you about future planned publications or any changes to the registry.

Key points:

- Freely available registry describing the *SAMD9/9L* mutations and associated phenotypes from investigators all over the world.
- Aggregated *SAMD9/9L* mutations and associated phenotypes will be displayed through the *SAMD9/9L* mutation database website.
- No patient-specific identifiable information will be collected.

If you are interested in participating in the registry or would like more information, please visit www.stjude.org/samd9. Link for data entry is available upon request.

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