the orphan disease center

MILLION DOLLAR BIKE RIDE
RAISING MONEY FOR RARE DISEASE RESEARCH

**ZC4H2** Associated Rare Disorders (ZARD) is an ultra-rare genetic condition with central and peripheral nervous system involvement caused by pathogenic variant of the ZC4H2 gene. ZC4H2 is located on the X chromosome and encodes the ZC4H2 (zinc finger C4H2-type containing) protein essential for normal development. ZARD can manifest in a broad range of clinical severity. Clinical presentations of affected individuals who carry the same pathogenic ZC4H2 gene variant can vary within families and between families. Males and females can be affected. To date, approx. 250 cases have been diagnosed worldwide.
There is currently very limited understanding of the function of the ZC4H2 gene and its protein. The focus for this grant opportunity is to a) understand the protein function of ZC4H2 and b) advance the search for viable therapies to treat this condition.

For this purpose, two equal grants of USD 46,846 each, will be offered to research projects on:

1. Studies on the function of ZC4H2 protein: Proteomic/transcriptomic profiling to identify protein function using patient-derived iPSCs (provided).
2. Novel therapeutic approaches for ZC4H2 Associated Rare Disorders (ZARD), including, but not limited to, X-reactivation, drug repurposing, techniques in genome editing, RNA-based mechanisms, biologics, novel cell-based therapeutics, and development of novel therapeutic compounds.

Applicants are expected to collaborate with other scientists and clinicians currently or previously involved in ZC4H2 research, and should include a statement on resource sharing in their proposal. Applicants are encouraged to use existing tools (e.g., existing viable and validated animal models, antibodies, fibroblasts, LCLs, iPSCs) and to contact the ZC4H2 Research Foundation (info@zc4h2foundation.com) with any questions about these resources. This grant is made possible by the ZC4H2 Research Foundation and the Orphan Disease Center.
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