MAST Genes Research Foundation
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Strategic Research Priorities

Originally approved by the board on 03/25/2023.
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Research Values

- Data produced during research should be shared openly to enable collaboration
- Research that is synergistic across the family of MAST genes is preferred over single-gene research
- Patients/caregivers should be directly involved in the research plan

Current Priorities

1. Build patient base (social media, registry) by engaging as many MAST families as possible
    - How many total patients?
    - From where (city/state/country)?
    - Which gene?
    - Which mutation?
    - Patient age and primary symptoms (related to #4 below)
    - Create partnerships with diagnostic tools and companies to connect patients/caregivers with the foundation for future research
2. Network and strategize with other rare disease groups (e.g. Project 8p, Jordan's Guardian Angels, Coalition to Cure CHD2, Syngap Research Fund)
3. Create a packet to send to families to share the long-term research plan and any short-term resources that could be helpful.

Future Priorities

1. Partner with other organizations (e.g. COMBINEDBrain, Rare Epilepsy Network, NORD, GeneDx, Invitae)
2. Explore drug repurposing studies
3. Develop questionnaire to inquire needs from families (e.g. which clinical symptoms most challenging; where would treatments have most impact for families)
    - Be able to look at needs of large group vs individual genes with the questionnaires.
    - Where are families located (city/state/country)
    - Which gene and mutation

    - Use questionnaire to establish priority areas for research
4. Look into biobanking (send which samples to whom? Ideally, in the US and contracted so Foundation can direct who/when samples can be used; Combined brain?).
5. Look into natural history options (e.g. Ciitizen, RareX, SPARK)
6. Host meetups for families to interact
7. Partner with American Epilepsy Society for grant
8. Establish a clinical center of excellence