From the lab to the clinic: An update on research & therapeutics

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Once upon a time.....

Two young adults, similar stories
Were there others?
YES!

• 14 patients
Gene discovery

- Gene (*WDR45*) mis-spelled in 13 of 14 patients!
- Gene makes beta-propeller protein
- “Beta-propeller protein-associated neurodegeneration”
- **BPAN**
Pathway to treatment

- Find the gene
- Build a patient registry and biosample library
- Make animal models
- Understand the disease
- Develop assays to test potential treatments
- Test drugs in animals
- Test drugs in people with BPAN
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Pathway to treatment

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Animal models
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Cells Constantly Recycle
Autophagy; Cells’ Recycle / Disposal System
Pathway to treatment

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Current projects

- Building the patient and family registry
- Developing assays to measure autophagy in BPAN
- Testing an FDA-approved drug in those assays
- Finding ways to measure BPAN
- Figuring out how to pay for it all
Lessons learned

Families are central
Power is in numbers
Collaboration is key
Funding is inadequate
Time is of the essence
Thank you!

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