



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

Penny Hogarth, MD

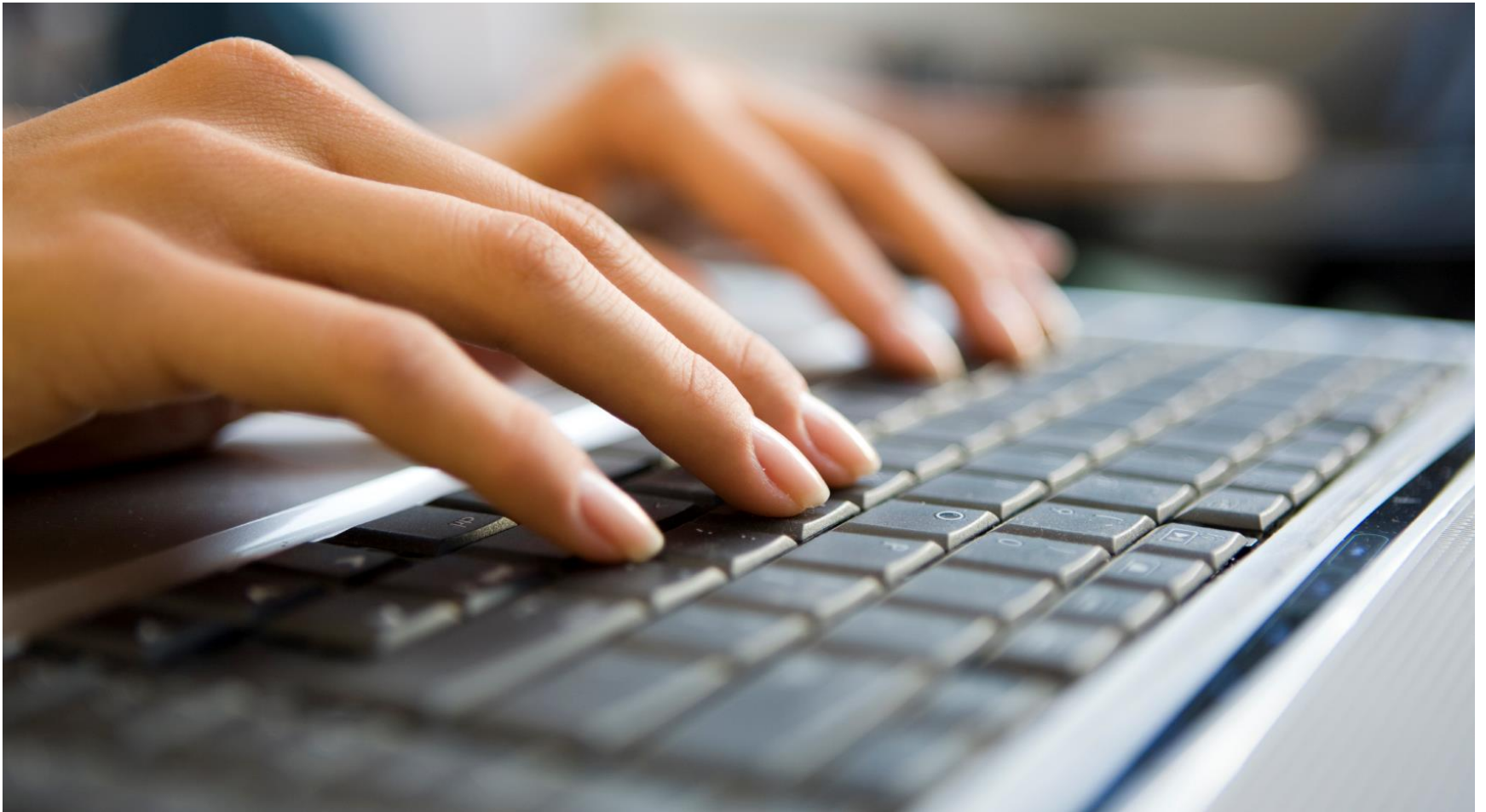
Oregon Health & Science University

Once upon a time.....

Two young adults, similar stories

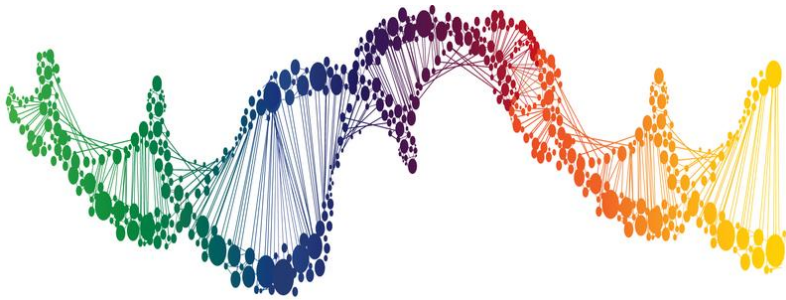


Were there others?



YES!

- 14 patients

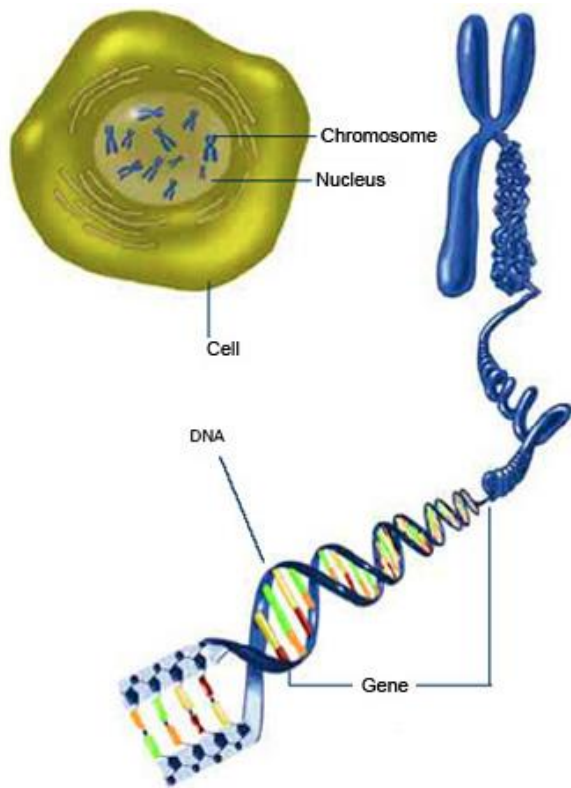


Gene discovery

- 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



NBIAcure

HOME ABOUT US LEARN OUR RESEARCH GET INVOLVED NEWS

DONATE



Welcome to NBIAcure

Although it may be out of reach today, a cure for each of the NBIA disorders is in our sights. We care deeply about improving the lives of children and adults with NBIA. Our team at NBIAcure will provide support and information to patients, families, clinicians and researchers, initiate new research studies to lay the foundation for clinical trials, and collaborate with academic and industry partners to develop new treatments.

We invite you to explore our site and return often to check on the latest news stories, find out about new studies that have launched, and learn how you can get involved. *Your participation is one of the keys to the cure.*

For the first time ever, people affected by an NBIA disorder can tell us about their disease and how it affects their lives. Better information about how each of these diseases progresses prepares us for therapeutic drug trials. We are building knowledge to determine if new drugs improve health and function. People with an NBIA disorder as well as family members will be able to contribute. *Stay connected here and help our community become 'trial ready.'*

Register

For Patients & Families

For Clinicians

For Researchers

NBIA DISORDERS

- PKAN
- PLAN (includes INAD)
- MPAN
- BPAN
- FAHN
- Other Disorders

OUR PARTNERS

- NBIA Family Community
- TIRCON

NBIAcure News



Announcement: 2016 BPAN Research Meeting!

BPAN research meeting

Our team is excited to announce that we will be hosting a BPAN research meeting in Portland, Oregon from June 24th-26th! We are thrilled to welcome you to our hometown and have planned a variety of activities including research activities, social events,...

REGISTER WITH US!
NBIAcure.org

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

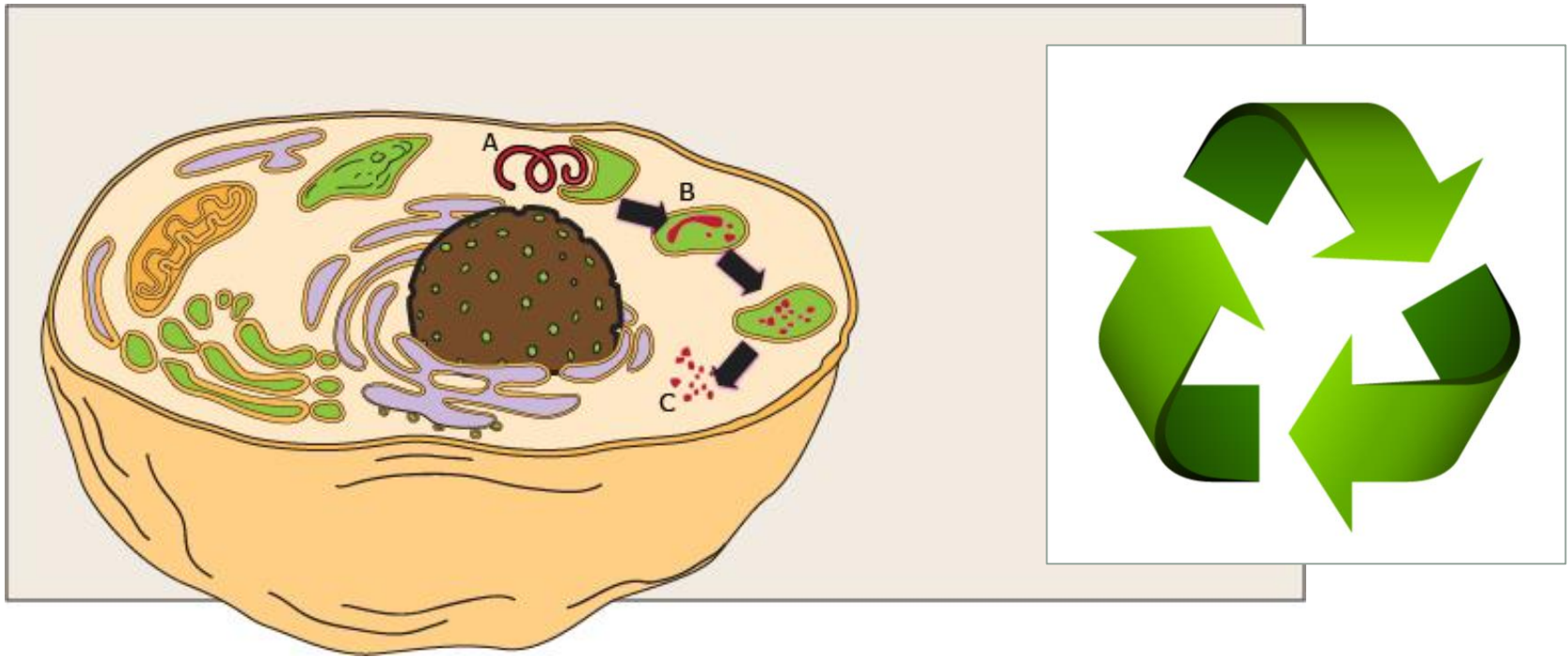
Animal models



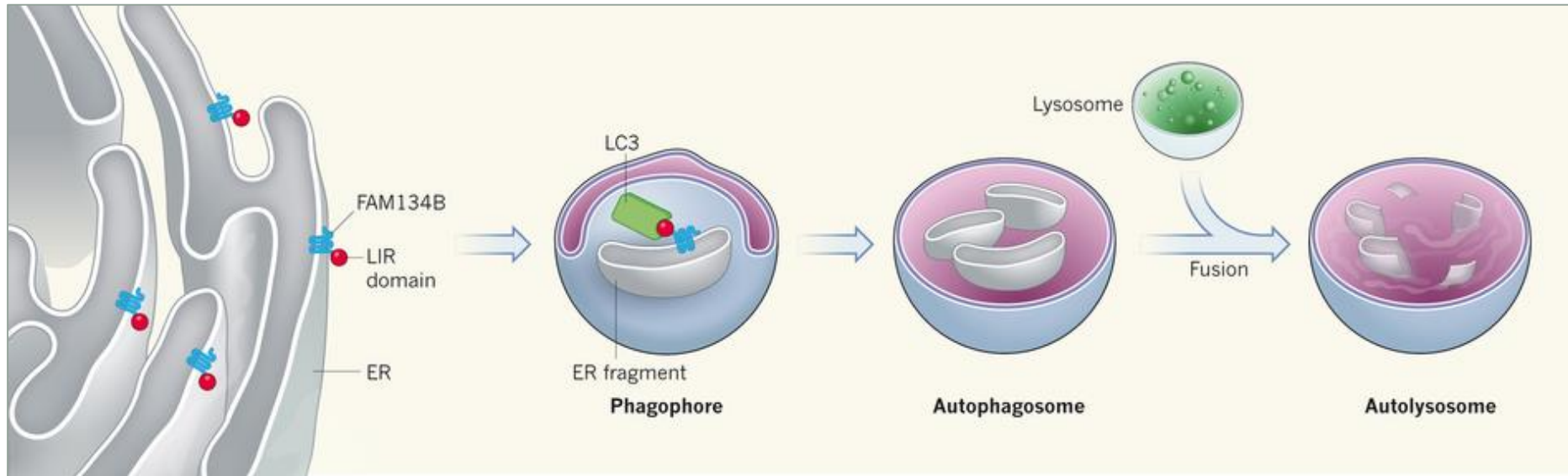
Pathway to treatment

- 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

Cells Constantly Recycle



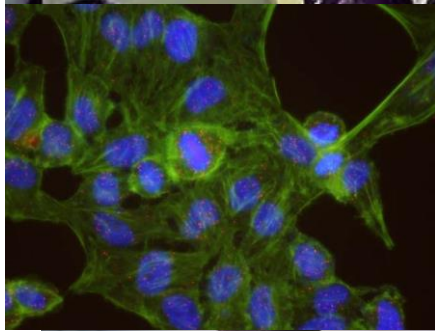
Autophagy; Cells' Recycle / Disposal System





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**



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!

- The Hayflick lab team:
 - **Susan Hayflick**
 - Allison Gregory
 - Leila Schwanneman
 - Puneet Rai
 - Suh Young Jeong
 - Rachel Fox
 - Randy Woltjer
 - Tanya Brooks
- Our volunteers and other staff
- Arletta
- Our funders & generous donors