

# WHAT WE HAVE LEARNED!

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DREAM IT.  
HOPE IT.

**National Ataxia Foundation**  
Annual Ataxia Conference  
April 1-3, 2016 ★ Orlando, Florida  
Hosted by the Southeast Region

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# PRESENTER DISCLOSURES

- Athena Diagnostics SCA5
- Consultant – Ionis and Biogen
- Research support – Ionis and Biogen, NIH, MDA



# FINDING A TREATMENT

- What do we need to develop a treatment for ataxia?
  - Organize Patients, Clinicians, Investigators, Pharma
  - Diagnose it
  - Understand it
  - Manage it optimally
  - Find a treatment
  - Evaluate treatment biology in patients
  - Validate treatment's clinical value
- How long will it take? – **at least 5 years from tomorrow**
  - Phase 1 – 1 year
  - Phase 2 – 1-2 years
  - Phase 3 – 2 years



# THE MAGIC OF A CURE



**But Other Types of Magic Require Skill,  
Education, and Hard Work**



# WHAT IS NEEDED TO MAKE AN ATAXIA TREATMENT POTION? REAL MAGIC OF ATAXIA TREATMENT

Management  
Management

Disease  
Disease  
Characterization  
Characterization

Diagnosis  
Diagnosis

Treatment  
Treatment  
Discovery  
Discovery

Treatment  
Treatment  
Evaluation  
Evaluation

Treatment Clinical  
Treatment Clinical  
Validation  
Validation



# IS THERE PROGRESS IN DIAGNOSIS

## MORE AND BETTER DIAGNOSES

### David Lynch

Importance of Genetic Dx of FA

### Vikram Shakkottai

Increasing ease of dx  
Increasing gene discovery

### Vikram Khurana

Importance of precise dx  
Clinical dx MSA and sporadic  
Gene dx

### Susan Perlman

### Chip Wilmot

Improved facility to dx  
Tailoring treatment to dx  
Focusing research on  
specific forms of ataxia



# DO WE UNDERSTAND THE BIOLOGY OF ATAXIA?

## CLEARER MECHANISMS OF ATAXIA

### David Lynch

Effects of frataxin loss on mitochondria and energy prod.

### Gülin Öz

160 investigators finding ataxia causes  
Clarifying causes in cells, animals and people

### Vikram Khurana

Connection of MSA and PD  
Role of protein folding in ataxia



### Henry Paulson and Lauren Moore

Stem cells to understand ataxia  
Using stem cells to test treatment

### Harry Orr

NAF helps new investigators  
NAF promotes new Rx

### Chip Wilmot

Common elements of SCA –  
protein folding  
nerve cell calcium loading  
RNA processing



# IS THERE PROGRESS IN CARE?

## OPTIMIZING ATAXIA MANAGEMENT

### Ellen Sichel

Importance of riding the wave  
Not letting ataxia have control

### Jonathan Rodis and Kathleen Kane

Navigating Disability  
SSI, SSDI and not giving up  
The importance of exercise –  
control balance to control gait

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### Nygel Lenz

Learning to honestly and  
openly deal with ataxia  
Fight ataxia, accept yourself

### Nancy Harrington

Novel communication means

### Susan Perlman

Control what you can control  
Sleep  
Exercise  
Nutrition – value of a feeding  
tube



# ARE THERE PROMISES OF TREATMENTS?

## REAL TREATMENTS ARE TAKING SHAPE

### David Lynch

Gene replacement therapy  
Energy production

### Hank Paulson and Lauren Moore

Need of research before stem  
cells can help with ataxia  
Power of ASOs

### Chip Wilmot

Marked increase in treatment  
discovery  
Increasing ease of dx  
Increasing gene discovery

### Vikram Khurana

Connection of MSA and PD  
opens treatment options  
Role of protein folding

### Gülin Öz

International ataxia  
investigators working  
together  
Many evolving treatments –  
new and repurposed drugs



# CAN WE TELL IF TREATMENTS WORK?

## BETTER QUANTIFICATION OF ATAXIA

### David Lynch

Clinical scales, biomarkers and tissue repository in FA

### Hank Paulson and Lauren Moore

BioBank for BioMarkers

### Chip Wilmot

Role of ataxia scales  
Tissue repository

### Jennifer Millar

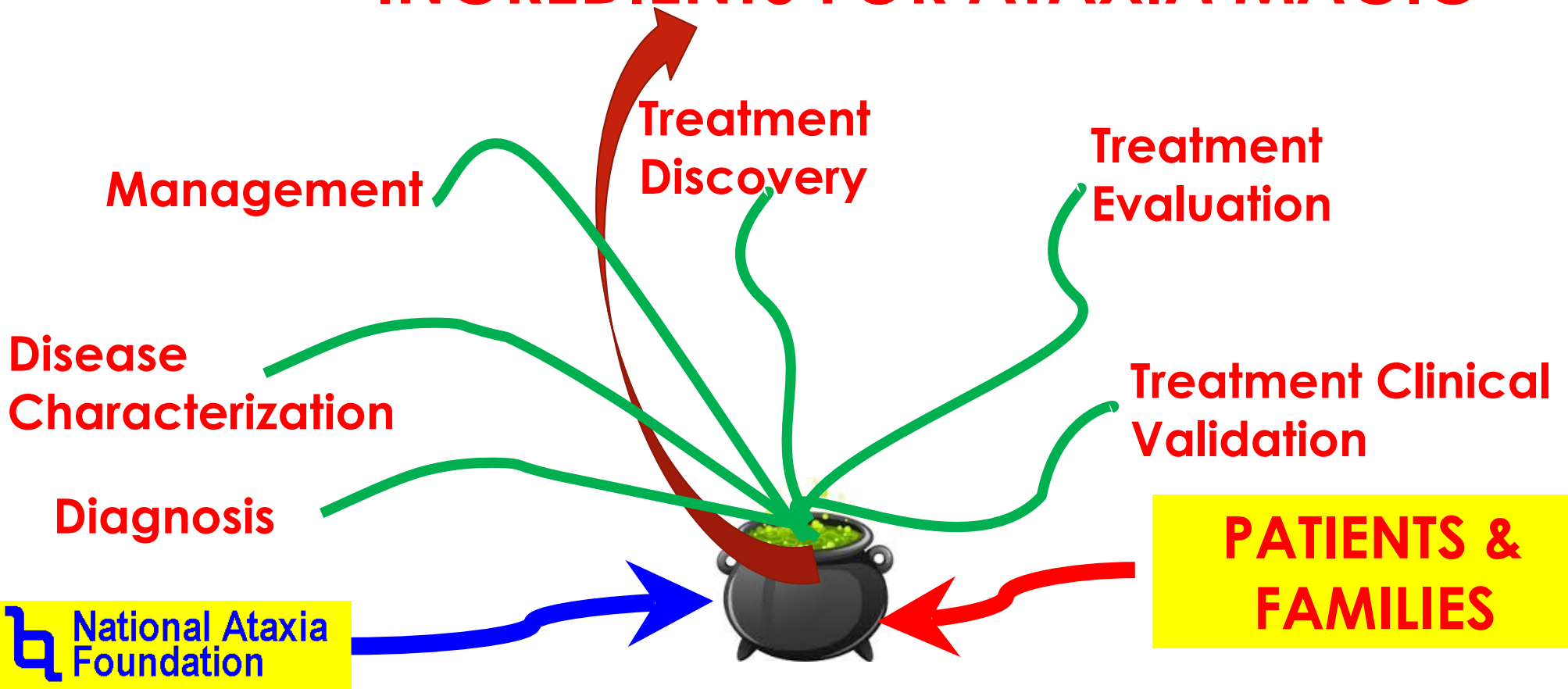
Gait lab to measure ataxia  
Quantitating motor function

### Gülin Öz

MRI measures of brain damage  
MRS early measures of disease  
Correlating MRI and Neuropsych



# INGREDIENTS FOR ATAXIA MAGIC



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**PATIENTS & FAMILIES**



THE MAGIC   
OF A CURE

## TAKE HOME MESSAGES

### Yes, Ataxia is Complicated

- Ataxia results from a large number of causes
- Each type of ataxia has unique damaging effects on nerve cells
- Each person with ataxia will need a team of sophisticated clinicians

### **BUT, There are more reasons than ever to be optimistic**

- Ataxia diagnosis and management are more standardized than ever
- Basic and clinical scientists have teamed-up to conquer ataxia
- Promising treatments are in the pipeline
- There is a light at the end of the tunnel – join the NAF to work toward