WHAT WE HAVE LEARNED!

John W. Day, MD, PhD
Professor of Neurology and Pediatrics – Stanford University
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PRESENTER DISCLOSURES

- Athena Diagnostics SCA5
- Consultant – Ionis and Biogen
- Research support – Ionis and Biogen, NIH, MDA
• 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
But Other Types of Magic Require Skill, Education, and Hard Work
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
Improved facility to dx
Tailoring treatment to dx
Focusing research on specific forms of ataxia

Chip Wilmot

National Ataxia Foundation
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

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IS THERE PROGRESS IN CARE?

OPTIMIZING ATAXIA MANAGEMENT

Ellen Sichel
Importance of riding the wave
Not letting ataxia have control

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

Nancy Harrington
Novel communication means

Jonathan Rodis and Kathleen Kane
Jennifer Millar
Navigating Disability
SSI, SSDI and not giving up
The importance of exercise – control balance to control gait

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

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ARE THERE PROMISES OF TREATMENTS?

REAL TREATMENTS ARE TAKING SHAPE

David Lynch
Gene replacement therapy
Energy production

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

Hank Paulson and Lauren Moore
Need of research before stem cells can help with ataxia
Power of ASOs

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

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CAN WE TELL IF TREATMENTS WORK?

BETTER QUANTIFICATION OF ATAXIA

David Lynch
Clinical scales, biomarkers and tissue repository in FA

Chip Wilmot
Role of ataxia scales
Tissue repository

Jennifer Millar
Gait lab to measure ataxia
Quantitating motor function

Hank Paulson and Lauren Moore
BioBank for BioMarkers

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

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INGREDIENTS FOR ATAXIA MAGIC

Management  
Disease Characterization  
Diagnosis  
Treatment Discovery  
Treatment Evaluation  
Treatment Clinical Validation  
PATIENTS & FAMILIES
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