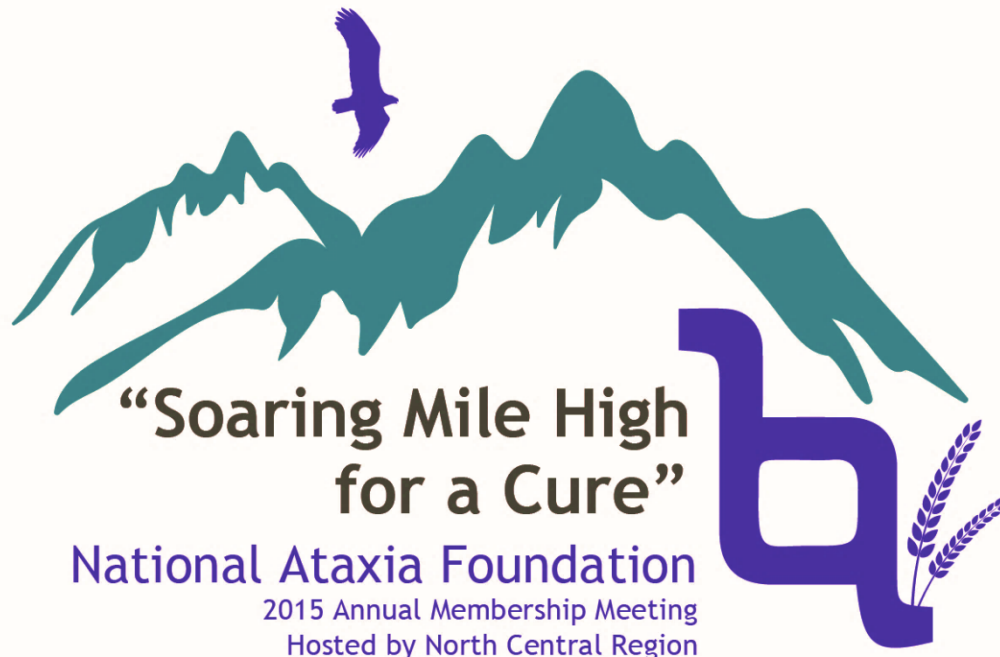


Highlights from the 2015 Annual Membership Meeting

Brent Fogel MD, PhD
Assistant Professor of Neurology
Program in Neurogenetics
University of California, Los Angeles



Disclaimer

- The information provided by speakers in any presentation made as part of the 2015 NAF Annual Membership Meeting is for informational use only.
- NAF encourages all attendees to consult with their primary care provider, neurologist, or other health care provider about any advice, exercise, therapies, medication, treatment, nutritional supplement, or regimen that may have been mentioned as part of any presentation.
- Products or services mentioned during these presentations does not imply endorsement by NAF.

Disclosure

- I, Brent Fogel, have no personal financial relationships with commercial interests relevant to this presentation within the past 12 months to disclose.



Highlights from Friday General Sessions



Michael Parent – NAF Update

- New Meeting records: More than 40% first time attendees & up to 650 people per meeting!
 - 1st meeting 1958 with 9 attendees
- Set New Records for Research in 2014
107 applications 19 countries → 24 funded
 - 1st grant \$5000 in 1978
 - Past 4 years nearly \$4,000,000 awarded!
- 2014 Membership Update
 - 3 chapters & 41 support groups

Temperature in Denver Friday Morning = 39° F

Temperature in Auditorium = 7° F (estimated)

Laura Ranum, PhD – NAF Research Update

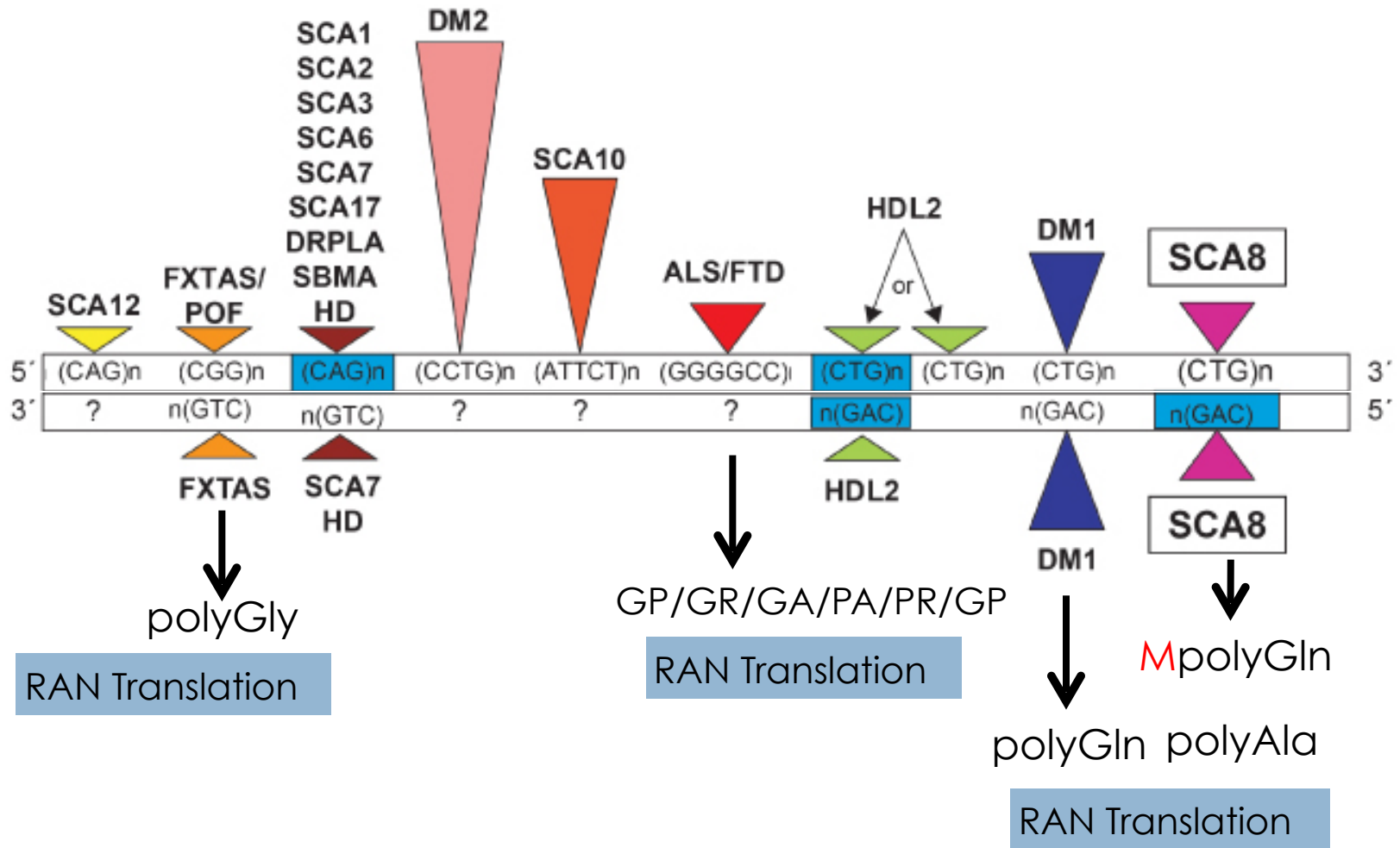
- *Genetics 101* → 2015



GENETICS

This is how it works

New view of expansion disorders



Framework for understanding disease mutations has changed
 Bidirectional expression is common
 Repeats express unexpected proteins
 Impact on strategies to treat these diseases



Abigail Collins, MD

Becoming an Adult with Ataxia



Ataxia

Independence

Relationships

Work

Society

Summary

- ▶ Becoming an adult is hard
- ▶ Becoming an adult with ataxia is harder
- ▶ Laws and Regulations exist to provide same rights and opportunities, but these are not always ideal
- ▶ Search the web for your local resources
- ▶ Enlist the help of a case manager to find the most appropriate opportunities and resources



Resources vary locally



Denver Community Resources for Youth with Disabilities

2013

Developed For: The Office of Economic Development,
Division of Workforce Development Youth Services
Youth Transitions Grant
720-865-5700

SECTION 11: EQUIPMENT/SERVICE NEEDS/HOME ADAPTATIONS PAGE 48

- Digital Federal Credit Union- Access Loans
- Friends of Man
- Hear Now
- Home Builders Foundation
- International Hearing Dog
- Kids Mobility Network, Inc.
- MS Society- Colorado Chapter
- Rebuilding Together
- Telecommunications Equipment Distribution Program

SECTION 12: TRANSPORTATION

PAGE 51

Transportation Assistance

- American Red Cross
- RTD Access-A-Ride

Driving

- Performance Mobility
- Colorado Adaptive Driving Programs
- Division of Motor Vehicles- Oral Driver's License Testing
- Master Drive

SECTION 13: INDEPENDENT LIVING CENTERS

PAGE 53

- Association of Colorado Centers For Independent Living
- Mile High Independent Living Center

Resources

- ▶ ADA information: <http://www.ada.gov>
- ▶ ADA-PARC: http://centerondisability.org/ada_parc/index.php which studies disparities that exist even with ADA law
- ▶ <http://www.parentcenterhub.org/repository/foradults/> national organization with information about resources including links to local and state programs
- ▶ www.disability.gov
- ▶ <http://www.easterseals.com/our-programs/adult-services/>



Theresa Chase, MA ND RN

Relationships & Sexuality





Barriers to Getting What You Want

- ▶ Other's ideas about sexuality & you....
- ▶ Your own ideas about sexuality & you.....
- ▶ Interference of the disability issues
- ▶ Changes in roles, relationship, daily life....
- ▶ Who's there? Single, partner, marriage, no one.
- ▶ Time, assistance, function
- ▶ Body Hatred & Body Image Issues

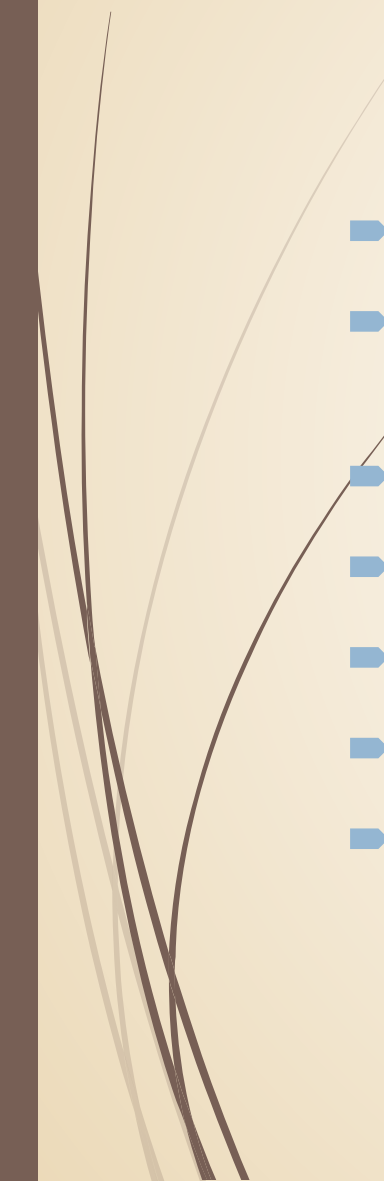


Expanding Your Views: Learning from Others

- ▶ People with Spinal Cord Injury Reported....
 - ▶ Increased sexual imagination
 - ▶ Knowledge & creativity
 - ▶ Discovered new erogenous zones
 - ▶ Explored new positions
 - ▶ Changed time of day routine
 - ▶ Planned sex was OK
 - ▶ Spasms enhanced sexual pleasure
 - ▶ Let go of the expectation of orgasm
- ▶ Experienced pleasure and orgasm in new and different ways.



What is Truly Great Sex?

- ▶ Being present, focused and embodied
 - ▶ Extraordinary communication and heightened empathy
 - ▶ Authenticity and transparency
 - ▶ Freedom to be genuinely honest
 - ▶ Feeling no self-consciousness with partner
 - ▶ Fun and Play....Sense of Humor...Laughing Together
 - ▶ Allowing oneself to be vulnerable and surrender
- 



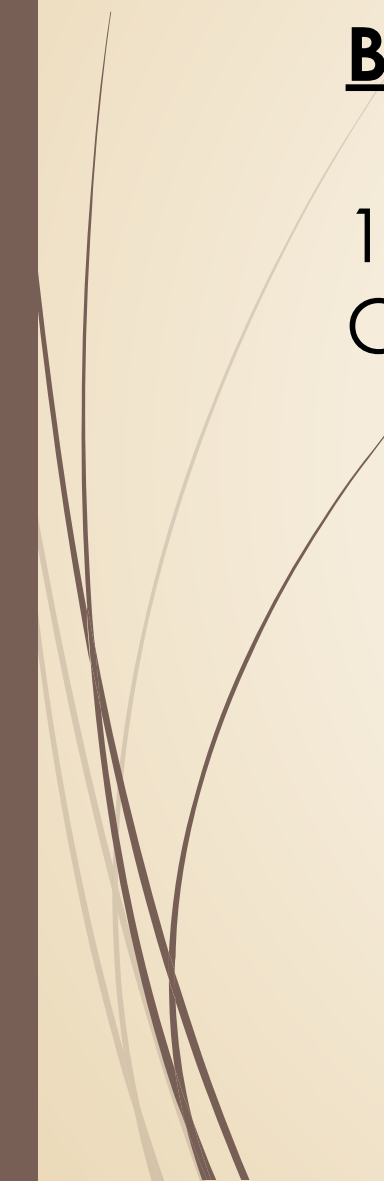
Highlights from Saturday General Sessions

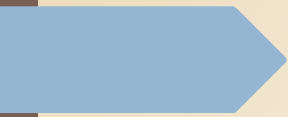
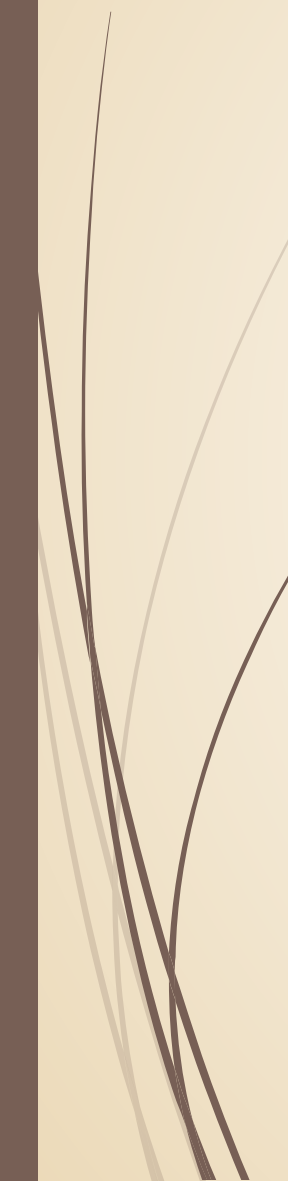




Bill Squires, CLU

10 Basic Financial Steps For Special Needs
Caregivers



- 
- 
- Step 1** Plan for the future
 - Step 2** Review Beneficiaries
 - Step 3** Family Meeting
 - Step 4** Create a Team
 - Step 5** Get Additional Resource Support
 - Step 6** Government Benefits
 - Step 7** Last Will and Testament
 - Step 8** Special Needs Trust
 - Step 9** Guardianship / Conservatorship
 - Step 10** The Letter of Intent

In summary

- Find quality professionals to assist you
- Remember that *YOU* are the expert about your dependent
- Think “lifetime care” and “quality of life”





Julie Hartman, PT DPT

Physical Therapy



Comprehensive Approach

Functional Training



Stability, Balance Coordination



Strength and Conditioning



Be Active!



Melissa Gibbons, MS CGC

Implications of Genetic Testing on
Families with Ataxia



Hurdles On the Path to Genetic Testing

**Provider Who Can Put it
All Together**

Insurance Coverage

Interpretation of Results



1

2

3


Impact of Family Planning

Reproductive Options

- ▶ Natural Conception
- ▶ Gamete (Sperm or Egg Donation)
- ▶ Preimplantation Genetic Diagnosis with In-Vitro Fertilization (IVF, ~\$20,000+)
- ▶ Prenatal Diagnosis
 - ▶ Chorionic Villus Sampling (CVS)
 - ▶ Amniocentesis
- ▶ Adoption



Resources

- ▶ Find a Genetic Counselor at **NSGC.ORG**
 - ▶ Find out more about GINA at **GINAHELP.ORG**
 - ▶ Does it Run in My Family Toolkit, Understanding Genetic and How To Talk to My Family about My Genetic Condition - **GENETICALLIANCE.ORG**
 - ▶ To learn more about all the ways that genetic is part of your life visit **GENESINLIFE.ORG**
- 



Michael Cammer, CPASG Co-Leader

My Journey with Ataxia





Michael Cammer: My Journey with Ataxia

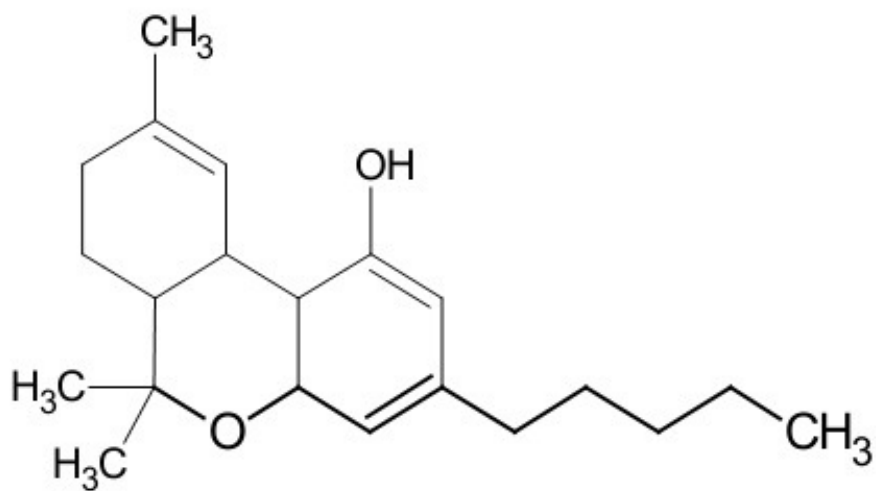
- ▶ You are not alone!
- ▶ You have all taken the first steps just by being here!
- ▶ Tell at least one person a day about Ataxia!
- ▶ Ataxians fall down enough on our own, we cannot let it keep us down!!!
- ▶ DON'T BE SHY!!!
- ▶ **Just one person can make a difference!**



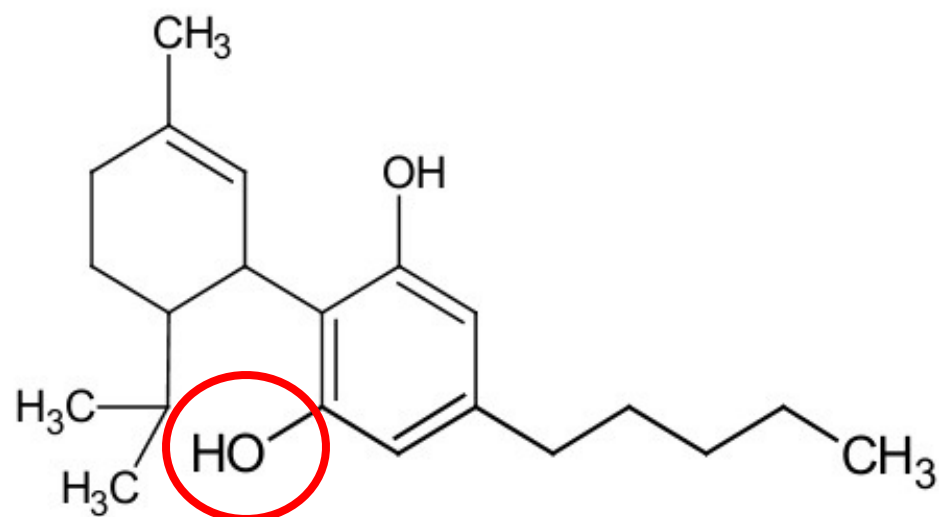
Terry Fife, MD

Medical Marijuana: A role in the Ataxias?





Tetrahydrocannabinol (THC)
psychoactive

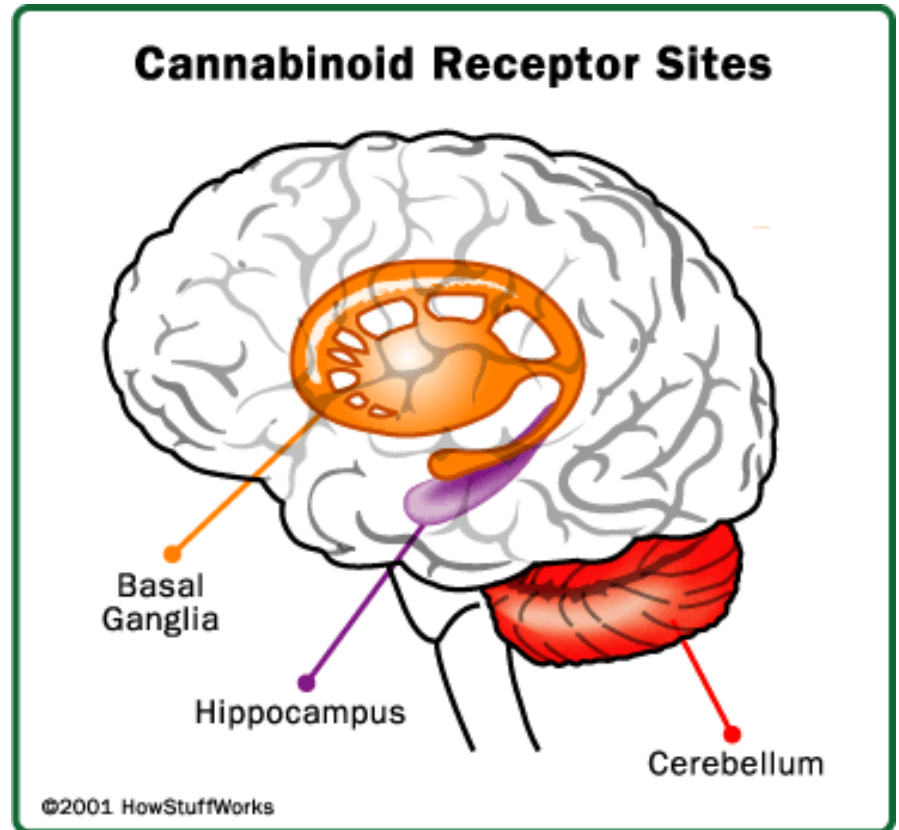


Cannabidiol (CBD)
not psychoactive

Brain has endocannabinoids which are our body's own molecules that bind to cannabinoid receptors (named CB1 and CB2).

CB1 – in CNS mainly hippocampus and cerebellum, basal ganglia, limbic system, prefrontal cortex.

CB2 – located on immune cells



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Special Article

Systematic review: Efficacy and safety of medical marijuana in selected neurologic disorders

Report of the Guideline Development Subcommittee of the American Academy of Neurology

Barbara S. Koppel, MD, FAAN, John C.M. Brust, MD, FAAN, Terry Fife, MD, FAAN, Jeff Bronstein, MD, PhD, Sarah Youssof, MD, Gary Gronseth, MD, FAAN and David Gloss, MD

Answers to Questions *preliminary*

1. *Refractory spasticity in patients with MS?*

Subjective improvement – YES

Objective improvement – NO

1. *Refractory central pain and painful spasms in MS?*

YES

2. *Bladder dysfunction in MS?* NO (mostly)

3. *Tremor in MS?* NO

4. *Dyskinesias of HD, dopa-dyskinesia in PD, Tics in Tourette's syndrome?* MOSTLY NO (possible modest reduction of chorea in HD)

5. *Seizure frequency in epilepsy?* NO

In need of more study before recommendation!



Highlights from Sunday General Sessions





Matt Feeney

Adaptive Recreation





THE STORY



JOEL BERMAN



MATT FEENEY

SPORTS
OPPORTUNITIES
• ALL

*The Mission of Adaptive Adventures is
To provide progressive outdoor sports opportunities to improve
quality of life for children, adults and veterans with physical
disabilities and their families*

SEASONS
• ALL
ABILITIES

ALL AGES

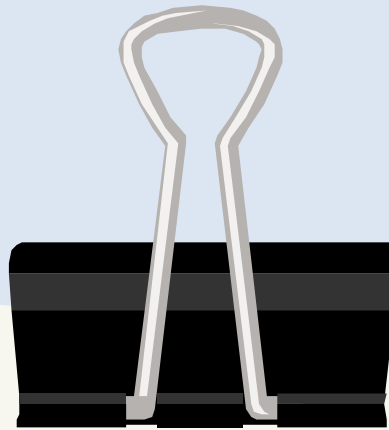




Naomi Hubert, MEd

Home Modifications, Adaptive
Technology and Accessible Housing





1.How to Complete a Home Assessment

2.Simple to Complex Home Mods

**3.Using Technology to Make Your Home
More Useable**

4.Advocating for Home Mods and AT

Solutions will be individual because each person with a disability is an individual

Different people facing a similar obstacle may require a different solution due to their specific needs

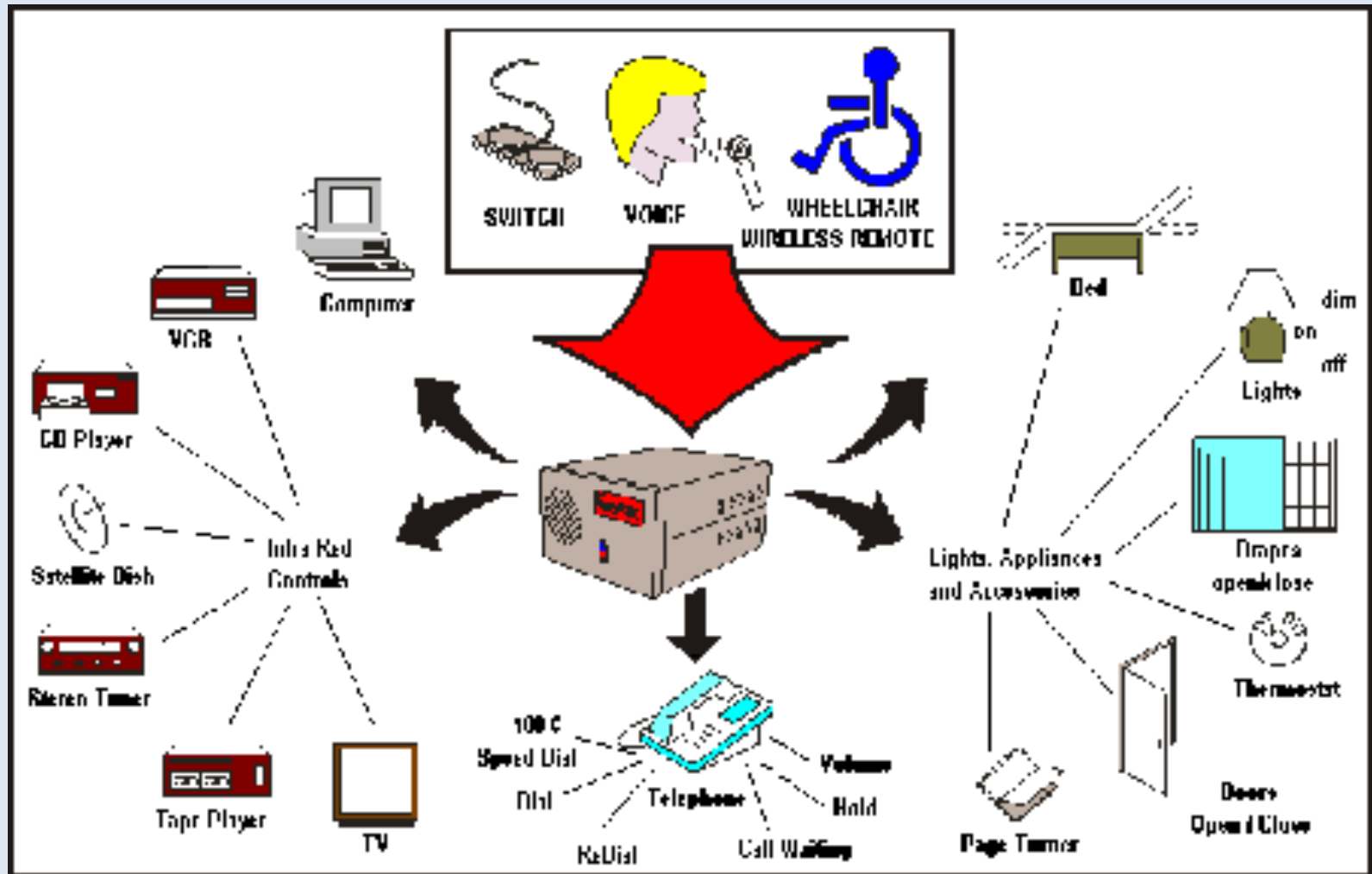




Fold Down Grab Bars

“Designer” Grab Bars

Environmental Control Units/ECU





Bailey Vernon, MPH CHES

Strategies for Living with Ataxia





Strategies

1. Execute a care plan
2. Exercise
3. Eat Well
4. Education
5. Effective Communication
6. Effective Coping
7. Exploring Options

Build Your Care Team

Patient

Family

Neurologist

Physical Therapist

Speech Therapist

Occupational Therapist

Nurse

Primary Physician

Dietician

Psychiatrist

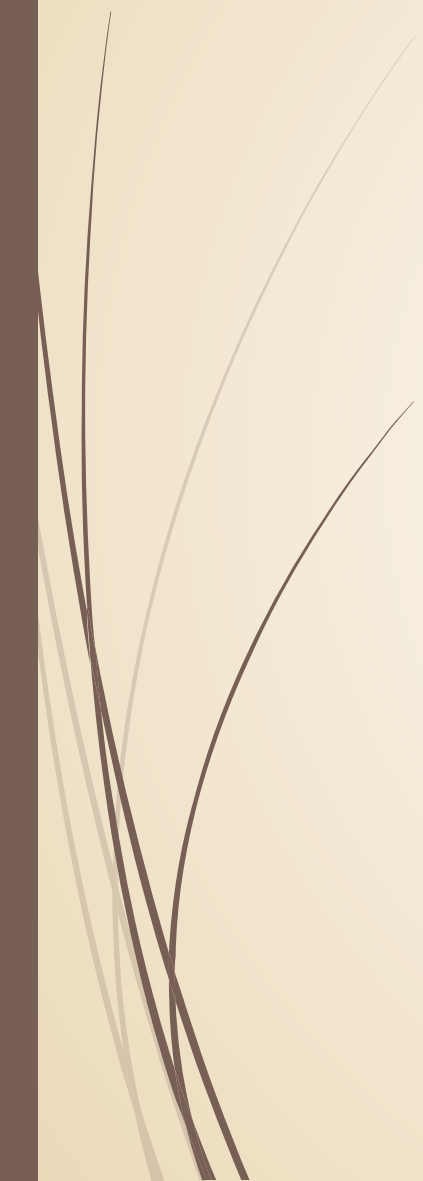
Genetic Counselor

Other Specialists

Support Groups



Effective Coping

- Stress management
 - Find the positive
 - Develop and maintain your humor
 - Professional services
 - Know your limits and make adaptations
 - Social support
 - Support groups
- 

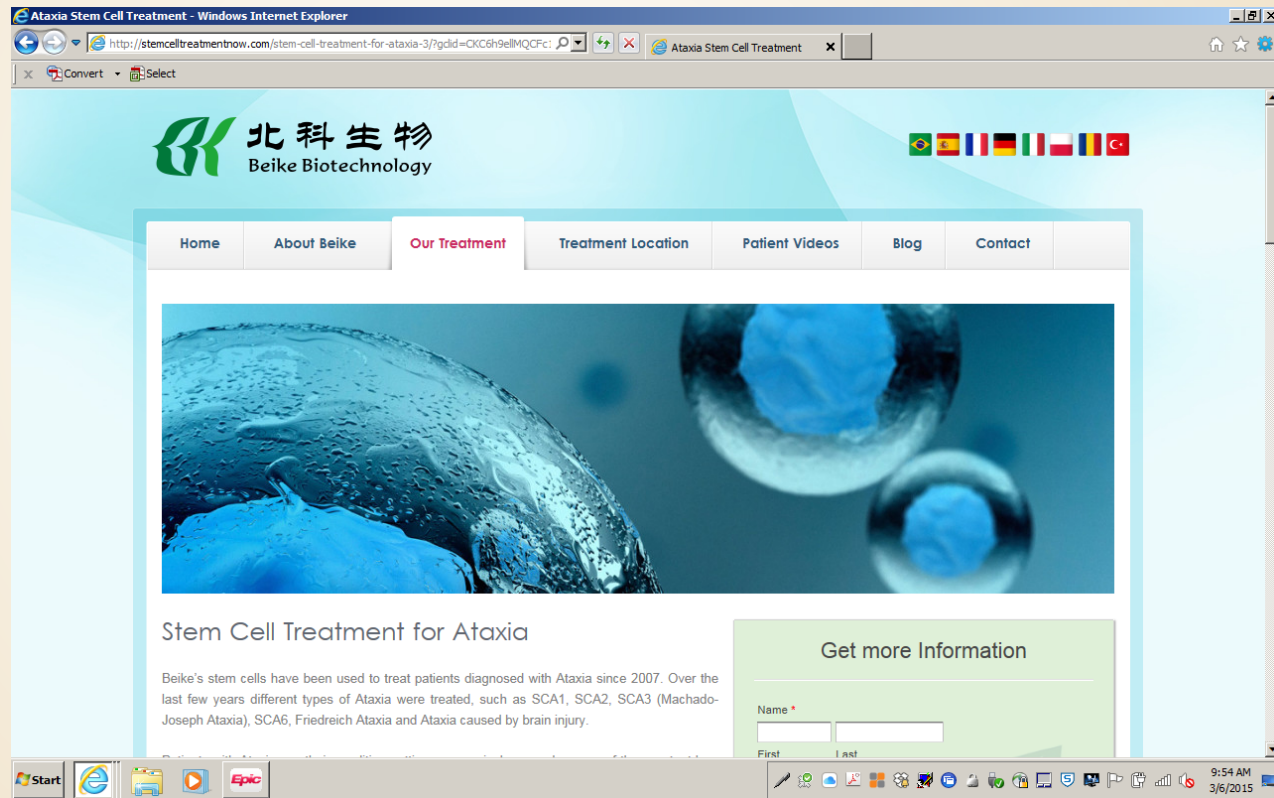


Susan Perlman, MD

The Ataxias: Research in FRDA, SCA, MSA



However, no matter what you read on the Internet...
there is still no cure for ataxia



Every Ataxia Patient Must Participate in Research/Clinical Trials

Registries

- ▶ For all ataxias--<https://www.sanfordresearch.org/CoRDS/CoRDSRegistryForm>
- ▶ For episodic ataxia--<http://rarediseasesnetwork.epi.usf.edu/cinch/index.html>
- ▶ For Friedreich's Ataxia--<http://www.curefa.org/registry.html>

Research in Ataxia

- **Human Subject Research**

- natural history studies
- biomarker studies for disease onset and progression
- mechanisms of degeneration

- **Basic Science Research**

- animal models
- cellular models (including iPSCs)
- genetic/molecular/epigenetic/environmental studies
- translational research

- **Clinical/Treatment Trials**

- *Since the AMM 2014, there have been 79 publications about ataxia treatment.*
- *Clinicaltrials.gov lists 57 open studies dealing with ataxia.*

SCA Pipeline—industry sponsored

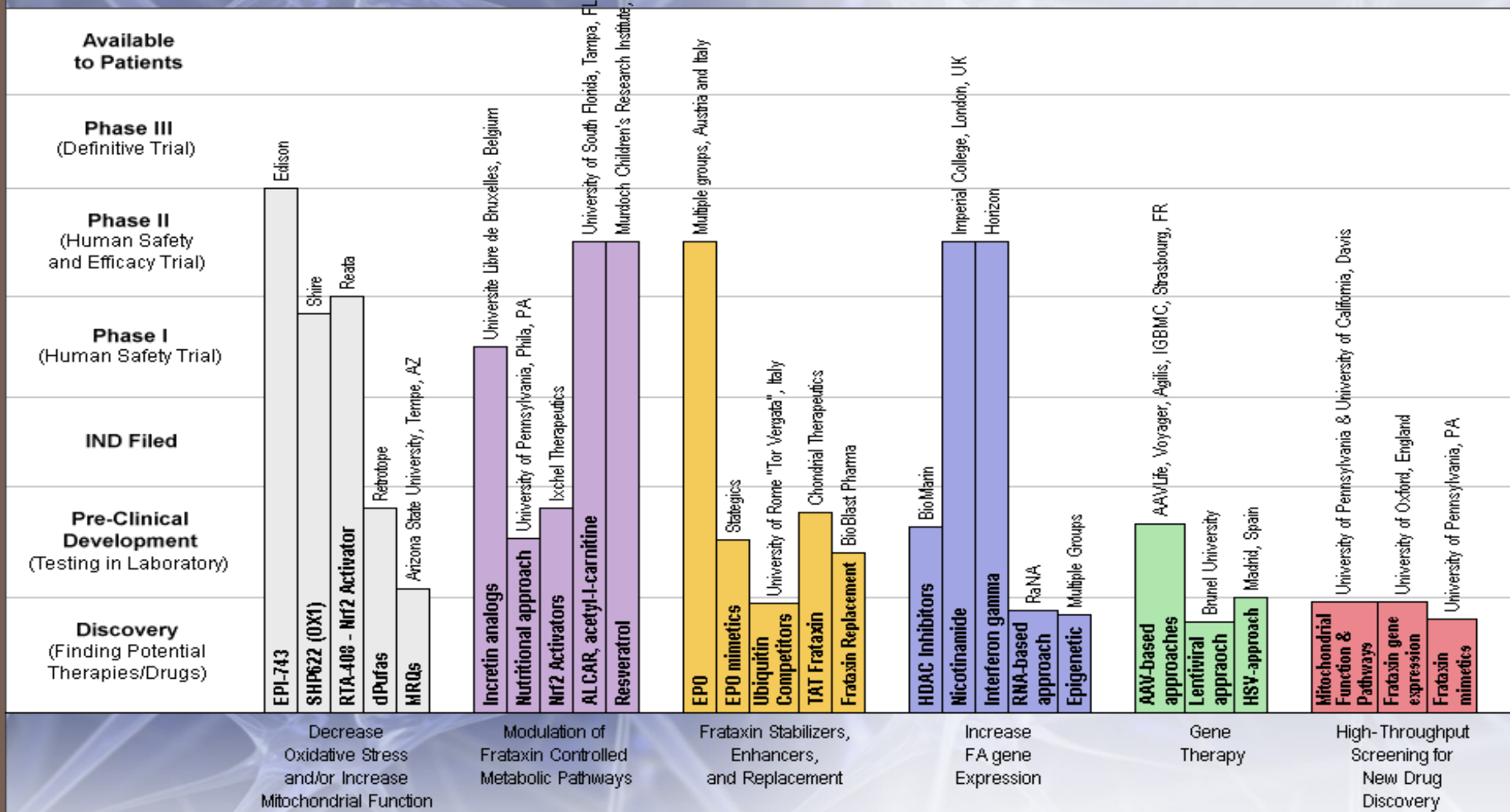
Company	Agent	MOA	Status
<i>Astra Zeneca</i>	<i>Myeloperoxidase inhibitor</i>	<i>Blocks microglial activation/ inflammation.</i>	<i>Phase 2 For MSA</i>
<i>Ataxion</i>	<i>CNS ion-channel modulators</i>	<i>Stabilizes Purkinje cell electrical potentials</i>	<i>Pre-Phase 1</i>
<i>Bioblast</i>	<i>Trehalose</i>	<i>Prevent aggregate formation</i>	<i>Phase 2 for SCA3</i>
<i>Isis Pharma</i>	<i>ASO ISIS-HTT</i>	<i>Antisense drugs bind to messenger RNAs (mRNAs) and inhibit the production of disease-causing proteins</i>	<i>Pre-Phase 1 For Huntington's disease</i>
<i>Prana Biotech</i>	<i>PBT2</i>	<i>Prevents the accumulation of toxic huntingtin proteins and the resultant damage inside neurons, and improve neuronal health and function by restoring normal copper and zinc levels which are disturbed when neurodegeneration takes place</i>	<i>Completed Phase 2 study in Huntington's disease.</i>

Friedreich's Ataxia Pipeline



Friedreich's Ataxia Research Alliance

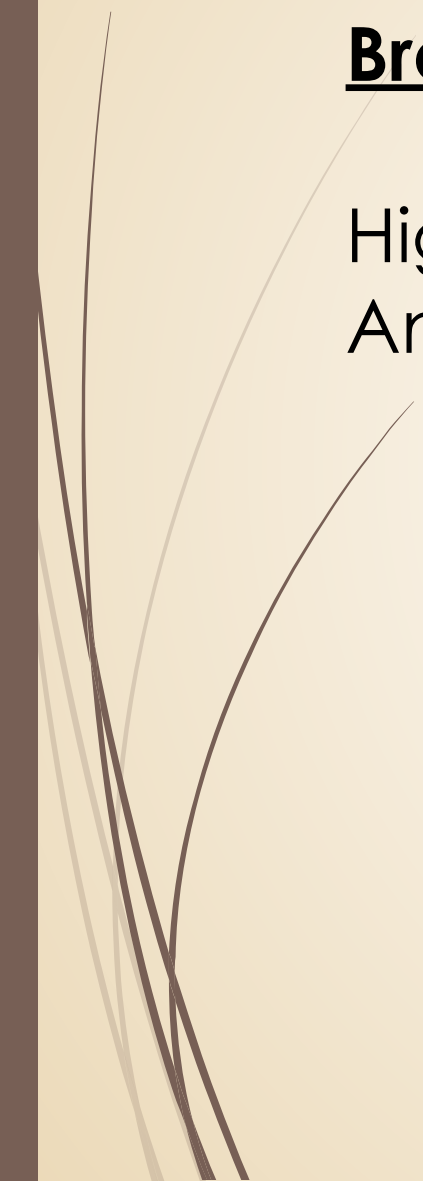
FA Treatment Pipeline December 2014





Brent Fogel, MD PhD

Highlights from the 2015
Annual Membership Meeting



This is **NOT** a highlight.

This is an angry cat.

This has nothing to do with ataxia.





Thank You!!!

