“Never doubt that a small group of thoughtful, committed citizens can change the world. Indeed, it is the only thing that ever has.”

Margaret Mead
Hannah Sames
Giant Axonal Neuropathy (GAN)
One of the most rare disorders in the world.
Timeline – scAAV9

- March 2008 – Diagnosis
- August 2008 – World’s First Symposium
- October 2008 – Began funding Dr. Gray
- 2010 Natural Hx Study began at UNC
  – Endpoints for Trials
- January 2012 – PreIND Mtg., FDA CBER
- December 2014 – NIH IRB approved the IND
- May 2015 – 1rst missense patient injected
- July 2016 – Hannah – 1rst null patient injected
Fundraising! - $6,000,000.00
Fundraising

• Two Doris Buffett matching challenge grants
• Two Pepsi Refresh Grants
• Two annual golf events with auctions
• Annual Gala with auctions
• Annual 5k
• Annual Texas Hold’em tournament
• “In-lieu of” and “In memory of” donations
• Ask people to make your charity, their charity of choice
Ground Zero – almost...

• Causal gene defect was known – recessive, loss-of-function disorder
• Mouse model was being developed
• Connected clinicians with scientists & neuropathologist evaluating the mouse
• Proof-of-concept/Dose Response/Tox. Studies
• GMP vector manufacture for human trials held at NIH under PI, Dr. Bonnemann
GAN - Monogenic

• Large phenotypic (symptomatic) variability
  – Kinky and straight hair cases
  – 6 yr olds using walkers & 16 yr olds who can jog
  – Siblings - 1 developed scoliosis and met all progression milestones 2 years earlier
  – Higher functioning kids with greater white matter disturbance than more progressed patients
  – 34 & 36 year old brothers who can drive their adaptive vehicle
22 years old
Advice

• Start a registry and get community sequenced
• Create a collaborative team with the shared goal of a Phase 1 trial – "CCTD" – Collab Coord Ther Dev
• Be focused – pick the therapeutic approach most likely to yield greatest patient benefit the fastest
• Fund a Natural Hx Study & connect MD with PhDs
• Align your disease with something “bigger”
• Avoid splinter organizations - use local campaigns
• Seek out industry partner and everyone must realize you are not the CF Foundation!
Thank you!
The Grey’s, in CA
Batten Disease (CLN2); 9 months from Dx to injection
Faces of GAN