“She has this – it is really rare”

“They are probably the most severely afflicted among carriers of chromosome abnormalities. They maintain the frog like position observed in infants and are reduced to an entirely vegetative, bedridden life.”
• Numerous specialists
• Healing problems
• At 2 yr. – poor growth

• At 3 yr. – Neurologist
  Hearing
1989
Find other parents

The Chromosome 18 Registry & Research Society

Help individuals with chromosome 18 abnormalities overcome the obstacles they face so they might lead healthy and productive lives.

The Syndromes of Chromosome 18

- 18q-
- Tetrasomy 18p
- Ring 18
- 18p-
Parent, Advocate, Researcher
Jannine D. Cody, PhD

1991 – 1997
Graduate School of Biomedical Sciences
UTHSCSA

May 1997

The Chromosome 18 Registry
Accomplishing our mission through:

**Service**
- Conferences (US, Australia & Europe)
- Regional Get-togethers
- Website / listservs /Facebook etc.

**Education**
- Congressional Testimony – 2 years
- Appropriations report language – 10 years
- Member presentations to - Anyone!

**Research**
- The Chromosome 18 Clinical Research Center
- $5.1 Million in research funding
The Chromosome 18 Clinical Research Center

- 7 Investigators with % effort
- 12 volunteer investigators
- 8 staff
- > 400 participants
  - Genotyped w/ 2 Kb resolution
- 31 publications
Team 18

Partnership

The Registry
- Conferences
- Member communication
- "Lobbying"
- Fundraising

The Research Center
- Human Participants
- Bench research
- Clinical research
- Expert collaborators
- Data analysis
The Google Rules

From “What Would Google Do?” by Jeff Jarvis

- Customers are in charge
- People can find each other
- Mass market is dead, now mass of niche markets
- Markets are conversations
- Product scarcity no longer guarantees profits
- Market premium comes from collaboration
- Success is based on networks and platforms
- Ownership is no longer key
• **Customers are in charge**

  Increased connectivity:
  - Customers have choices
  - Locality not important
  - Instant speakers platform
  - Crowd (fans or enemies)
  - Companies that cede control win

• **People can find each other**

  Increased connectivity:
  - < 6 degrees of separation
  - People can stay in touch
    - More acquaintances
    - Longer period of time
  - Kindred spirits are easily found

• **The mass market is dead, now a mass of niche markets**

  Released from Atoms
  No longer important to predict the next big hit
  - Self publishing
  - Self recording
  - Self marketing
• *Markets are conversations*

Marketing is out and conversations are in

Old Model:
Professionals produced > amateurs consumed

New Model:
Let customers do the work and learn from it.

• *Product scarcity no longer guarantees profits*

  Increased connectivity:

  • The only store in town, but not the only store that is accessible.

  • Locality is not the key to success

• *Market premium comes from collaboration*

  Increased connectivity:

  • Open sourcing
    Britannica > Wikipedia
• Success is based on networks and platforms

• Ownership is no longer key

  Increased connectivity:

  • Goldcorp Challenge
  • Released geological data
    1996 - 53,000 oz @ $360/oz
    2001 - 504 oz @ $59/oz

The Google Rules
From “What Would Google Do?” by Jeff Jarvis

A fundamental change in the balance of power
What do the Google Rules mean to the Rare Disease Community?

Rare is no longer second class!

Rare ≠
  • Hard to find
  • Isolated
  • Without a collective voice
Rare =
  • Focused constituency
  • Informed participants

WWGD for the Rare Disease Research Community?

Phenotype rules! = genotyping is so last century

Phenotypes are not dichotomous
  Age of onset
  Severity
  Levels of X
  Associated findings
  Natural history (or unnatural history)

WWGD for the Rare Disease Research Community?

• Participant access to their own data
• Participants can update their own data
• Participants can query the data set to get anonymized data
• Participants have interactive platform (a private and secret Facebook page)
What Can This Mean for Researchers?

- You cede control
- Extend your eyes and ears
- Increased understanding means increased involvement

What Can This Mean for Advocates?

- Focused on the things that matter to families
- Better understanding of the condition
- Accelerate the race for a cure

The World Congress on Chromosome Abnormalities

June 2004
San Antonio, Texas
Us!

We have a long way to go!

R01 Summary Statements from June 2010

We have a long way to go!

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Recommended Reading

*What Would Google Do?* By Jeff Jarvis
*The World is Flat* by Thomas L. Friedman
*The Long Tail* by Chris Anderson
*The Black Swan* by Nassim Nicholas Taleb

With future paylines at 3%, you better read:
*Rich Dad Poor Dad* by Robert Kiyosaki and Sharon Lechter