

Parent, Advocate, Researcher
(the view from my parachute)

Founder & President

Associate Professor
Dept. of Pediatrics

Scientific Director
CHART Center

1985

- Cleft palate
- Metatarsus adductus
- Bright-eyed
- Alert

“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.”








1989
Find other parents

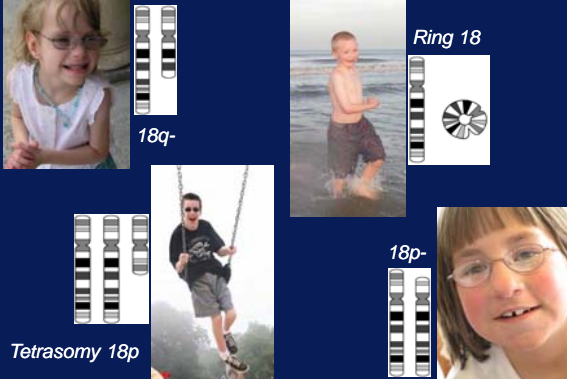


The Chromosome 18 Registry & Research Society



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

The Syndromes of Chromosome 18



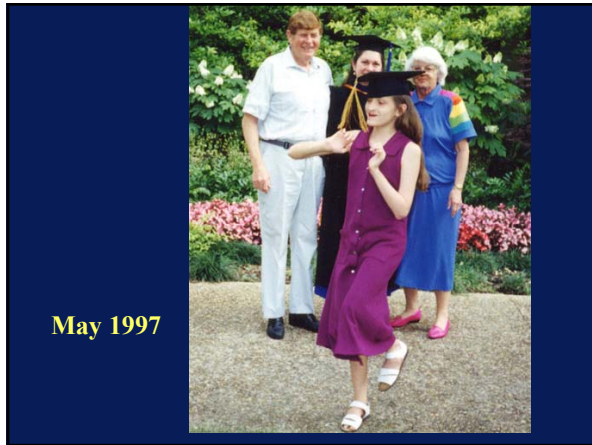
18q-

Ring 18

Tetrasomy 18p

18p-







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





UT HEALTH SCIENCE CENTER™
The Chromosome 18 Clinical Research Center

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





Partnership



<p><u>The Registry</u></p> <ul style="list-style-type: none">• Conferences• Member communication• “Lobbying”• Fundraising	<p><u>The Research Center</u></p> <ul style="list-style-type: none">• Human Participants• Bench research• Clinical research• Expert collaborators• Data analysis
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BMJ VOLUME 327 26-27 DECEMBER 2003 bmj.com Hazardous journeys

Parachute use to prevent death and major trauma related to gravitational challenge: systematic review of randomised controlled trials

Gordon C S Smith, Jill P Pell

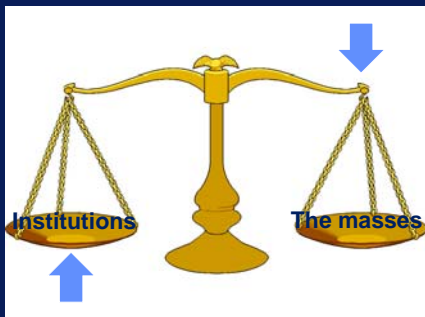
Abstract
Objectives To determine whether parachutes are effective in preventing major trauma related to gravitational challenge.
Design Systematic review of randomised controlled trials.
Data sources Medline, Web of Science, Embase, and the Cochrane Library databases, appropriate internet sites and citation lists.
Study selection Studies showing the effects of using a parachute during free fall.
Main outcome measures Death or major trauma, defined as an injury severity score > 15.
Results We were unable to identify any randomised controlled trials of parachute intervention.
Conclusions As with many interventions intended to prevent ill health, the effectiveness of parachutes has not been subjected to rigorous evaluation by using randomised controlled trials. Advocates of evidence based medicine have criticised the adoption of interventions evaluated by using only observational data. We think that everyone might benefit if the most radical protagonists of evidence based medicine organised and participated in a double blind, randomised, placebo controlled, crossover trial of the parachute.



Parachutes reduce the risk of injury after gravitational challenge, but their effectiveness has not been proved with randomised controlled trials

The Google Rules

From "What Would Google Do?" by Jeff Jarvis



A fundamental change in the balance of power.

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



A network diagram with a central blue box labeled "Acme Widgets" connected to several smaller purple boxes, representing a network structure.

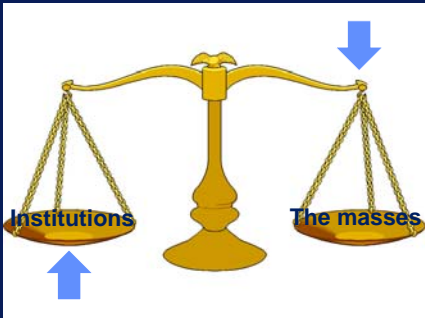
•Ownership is no longer key

Increased connectivity:

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

The Google Rules

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A golden scale of justice. The left pan is labeled "Institutions" and has a blue arrow pointing up towards it. The right pan is labeled "The masses" and has a blue arrow pointing down towards it.

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



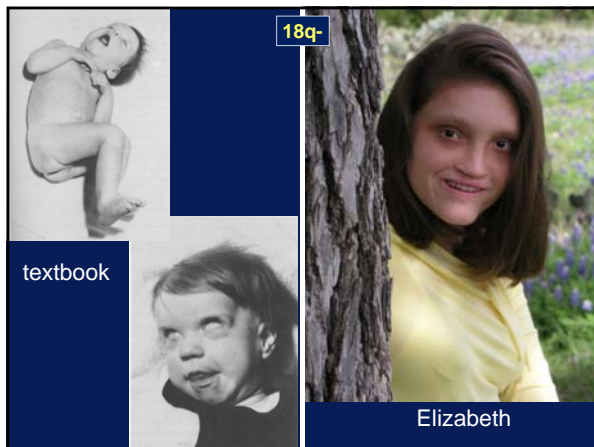
June 2004
San Antonio, Texas



We have a long way to go!

R01 Summary Statements from June 2010

Weaknesses <ul style="list-style-type: none">• Study involves only chromosome 18q disorders.
Weaknesses <ul style="list-style-type: none">• Current application is in a rare disorder
Weaknesses <ul style="list-style-type: none">• Not a randomized trial



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 Dan Poor Dad by Robert Kiyosaki and
Sharon Lechter
