How Research Can Benefit Families and Patients

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Research.....it’s a good thing!
WHY IS RESEARCH IMPORTANT?

“....essential for improving the quality of human lives”

Knowledge about health & disease
- Natural history of outcomes
- Causes of diseases
- Who is at risk
- Biological mechanisms

Develop and test treatments or approaches
- What works?
- What doesn’t work?
- Risks and benefits

Improve the health and wellbeing of people
- Prevent or cure disease
- Treat conditions
- Improve quality of life
Leo
Autism Prevalence On The Rise*
There has been a 600% increase in prevalence over the last two decades.

AUTISM SPEAKS*
It's time to listen.
www_AUTISM Speaks.org

*Recent research has indicated that changes in diagnostic practices may account for at least 25% of the increase in prevalence over time, however much of the increase is still unaccounted for and may be influenced by environmental factors.
From Denver.... to Early Start Denver Model
Autism
45,075 publications*

Source: US National Library of Medicine National Institutes of Health
Fragile X-Associated Tremor Ataxia

A Brief History of Advances in Fragile X Knowledge

- **1943**, Martin and Bell: described a pedigree of X-linked mental disability 26 years later.....
- **1969**, Lubs: "marker X chromosome,"chromosomal test for Fragile X,
- **1970**, Hecht: coined "fragile site".
- **1970-1980's** cytogenetic diagnostic test for Fragile X syndrome
- **1985**, de la Cruz: outlined physical, psychological, & cytogenic characteristics.
- **1990's**, 2 molecular DNA tests (Southern Blot/Polymerase Chain Reaction (PCR))
- **1991**, the FMR1 gene and mutational basis of Fragile X syndrome discovered.
- **1994**, AGG interruptions found to affect stability of the Fragile X Triplet repeat
- **2001**, Initial cases of FXTAS and FXPOI were recognized
- **2010**, AGG testing commercially universal
- **2010**, FMRP a highly efficient PCR-based assay developed
- **2018**, North Carolina launches Newborn screening pilot, Early Check
- **2018**, FXAND was first described***
Fragile X Syndrome
6,261 Publications*

*Source: US National Library of Medicine National Institutes of Health
What can just one research paper do?

A review of trisomy X (47,XXX)

Nicole R Tartaglia, Susan Howell, Ashley Sutherland, Rebecca Wilson and Lennie Wilson

Orphanet Journal of Rare Diseases 2010 5:8
https://doi.org/10.1186/1750-1172-5-8 © Tartaglia et al; licensee BioMed Central Ltd. 2010
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Abstract

Trisomy X is a sex chromosome anomaly with a variable phenotype caused by the

36 participants led to international impact
X and Y Variations......
Google…

- Autism: 198,000,000 results
- Fragile X Syndrome: 13,600,000 results
- XXY Syndrome: 499,000 results
- XYY Syndrome: 182,000 results
- XXX Syndrome: 11,600 results
- XXXY Syndrome: 6,720 results
- XXYY Syndrome: 5,700 results
1. Access
2. Contribute
3. Advance knowledge