

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*

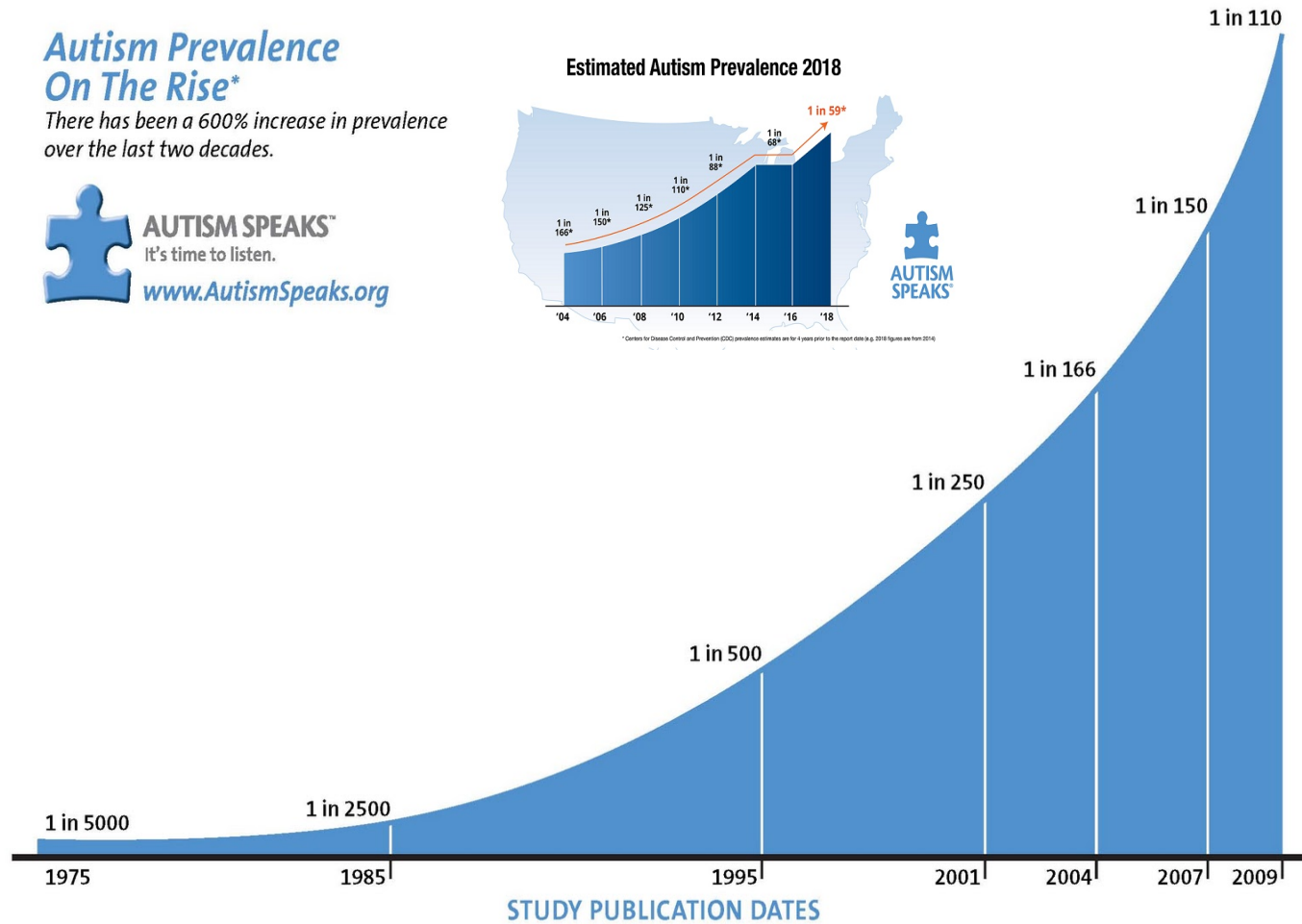
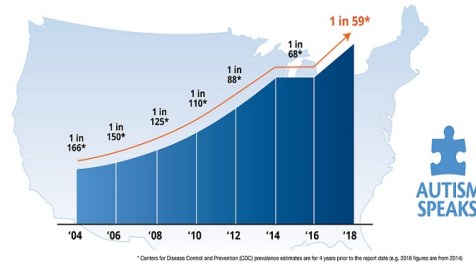
There has been a 600% increase in prevalence over the last two decades.



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www.AutismSpeaks.org

Estimated Autism Prevalence 2018



*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.

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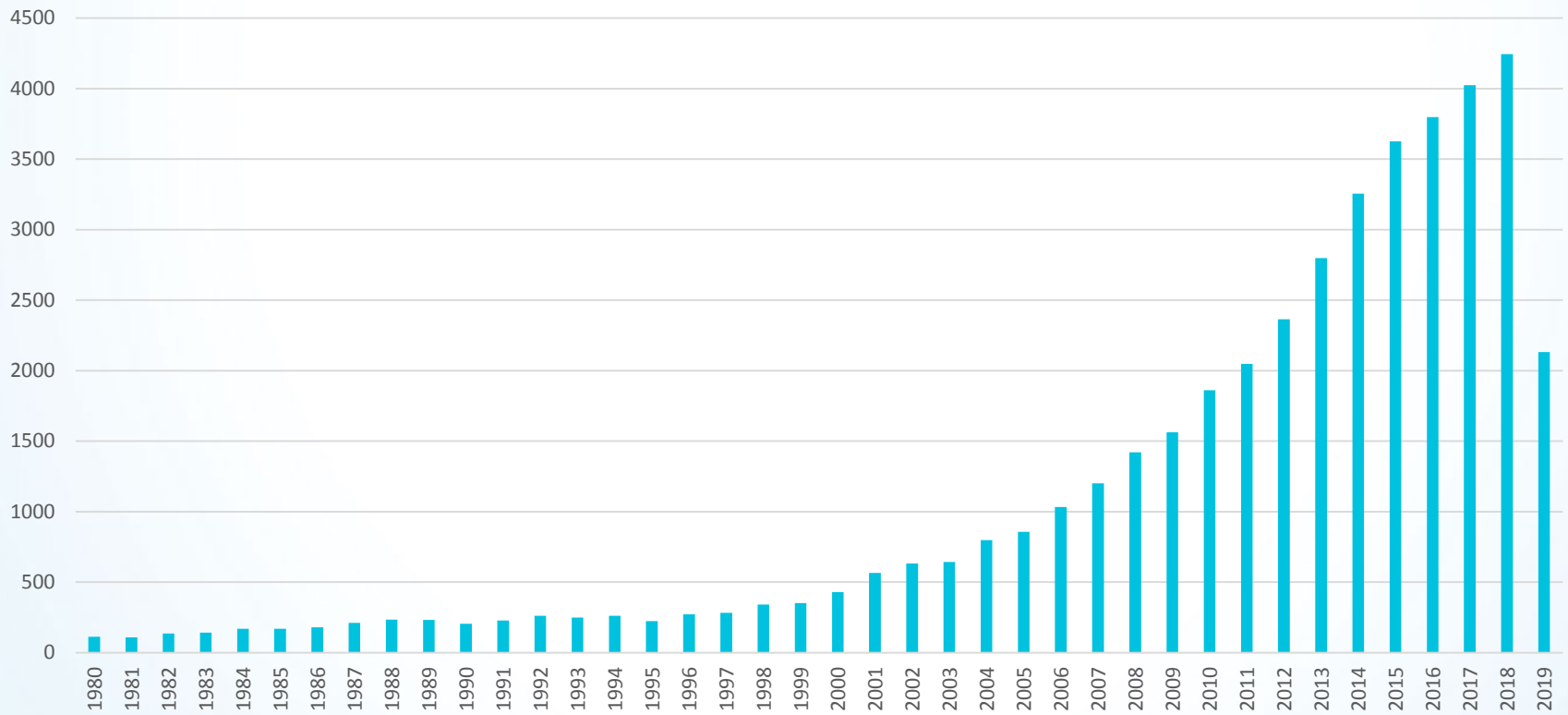


From Denver.... to Early Start Denver Model



Autism

45,075 publications*



[Source: US National Library of Medicine National Institutes of Health](#)



Fragile X-Associated Tremor Ataxia

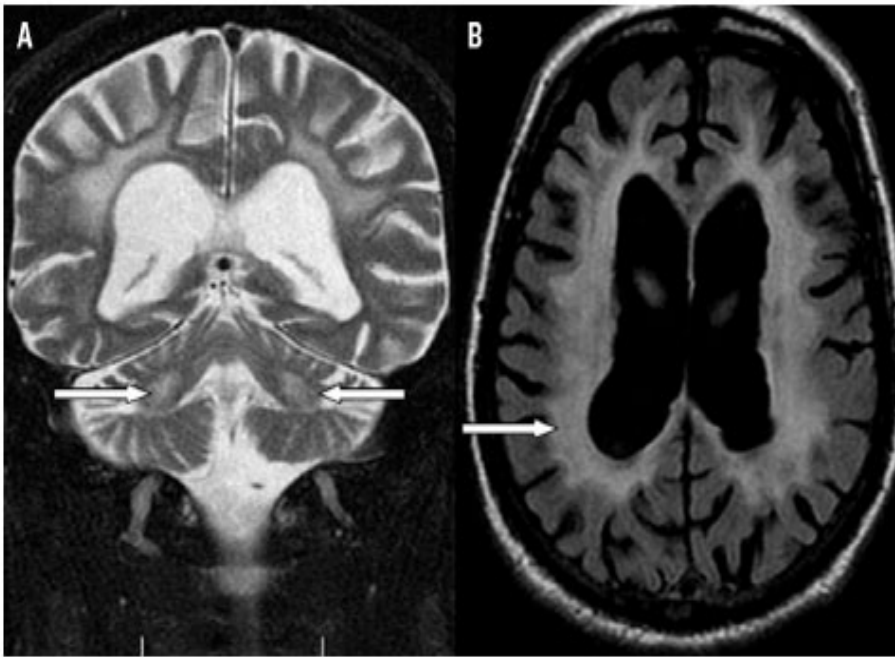


Figure. MRI features of a fragile X-associated tremor and ataxia syndrome patient: [A] increased T2 signal intensity (arrows) in white matter of the middle cerebellar peduncles; [B] increased signal intensity (arrow) on FLAIR images in periventricular and deep white matter of the hemispheres.

Source: Arq. Neuro-Psiquiatr. vol.68 no.5 São Paulo Oct. 2010, The fragile x-associated tremor and ataxia syndrome (FXTAS), Leonardo Pires Capelli; et al



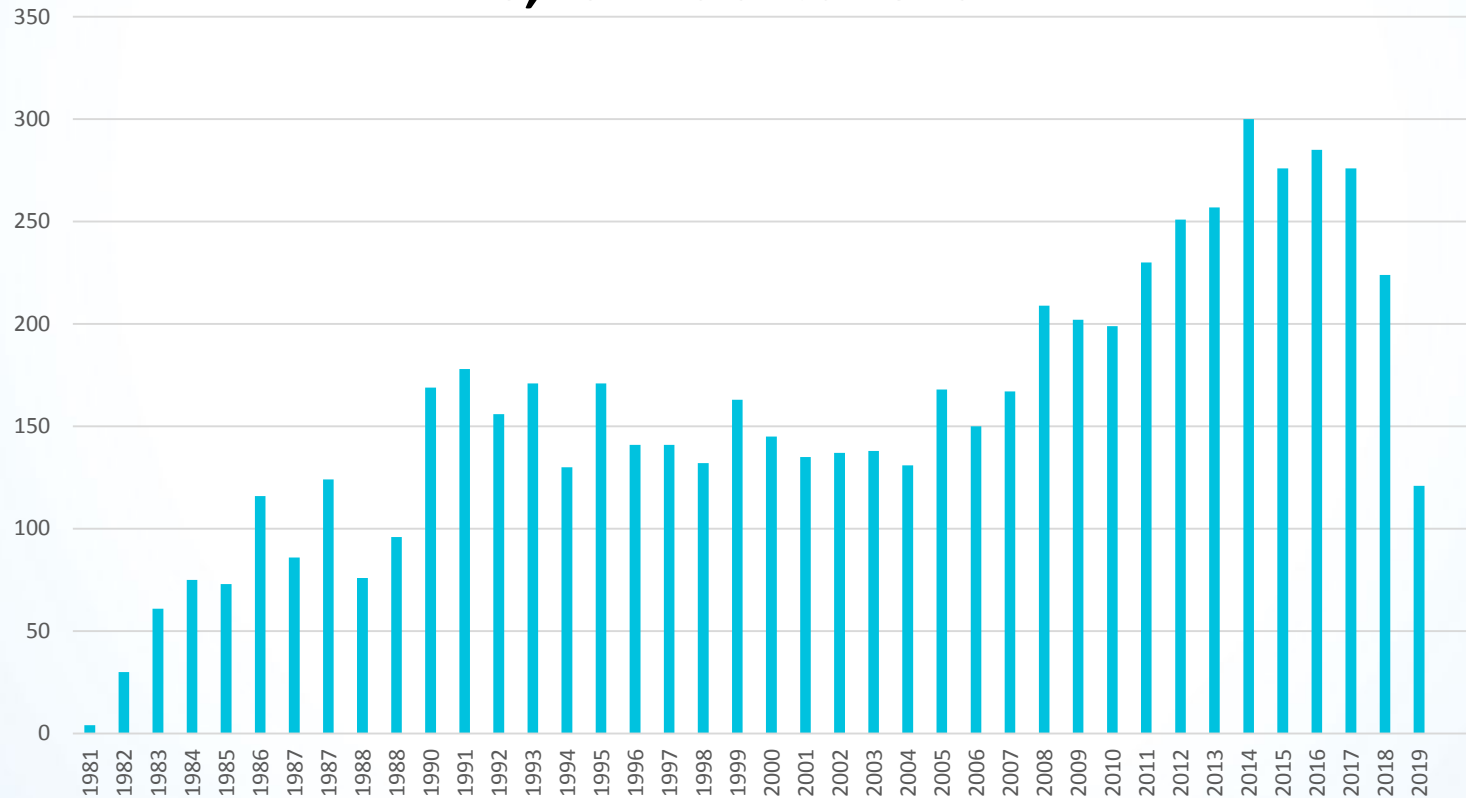


A Brief History of Advances in Fragile X Knowledge

- **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***

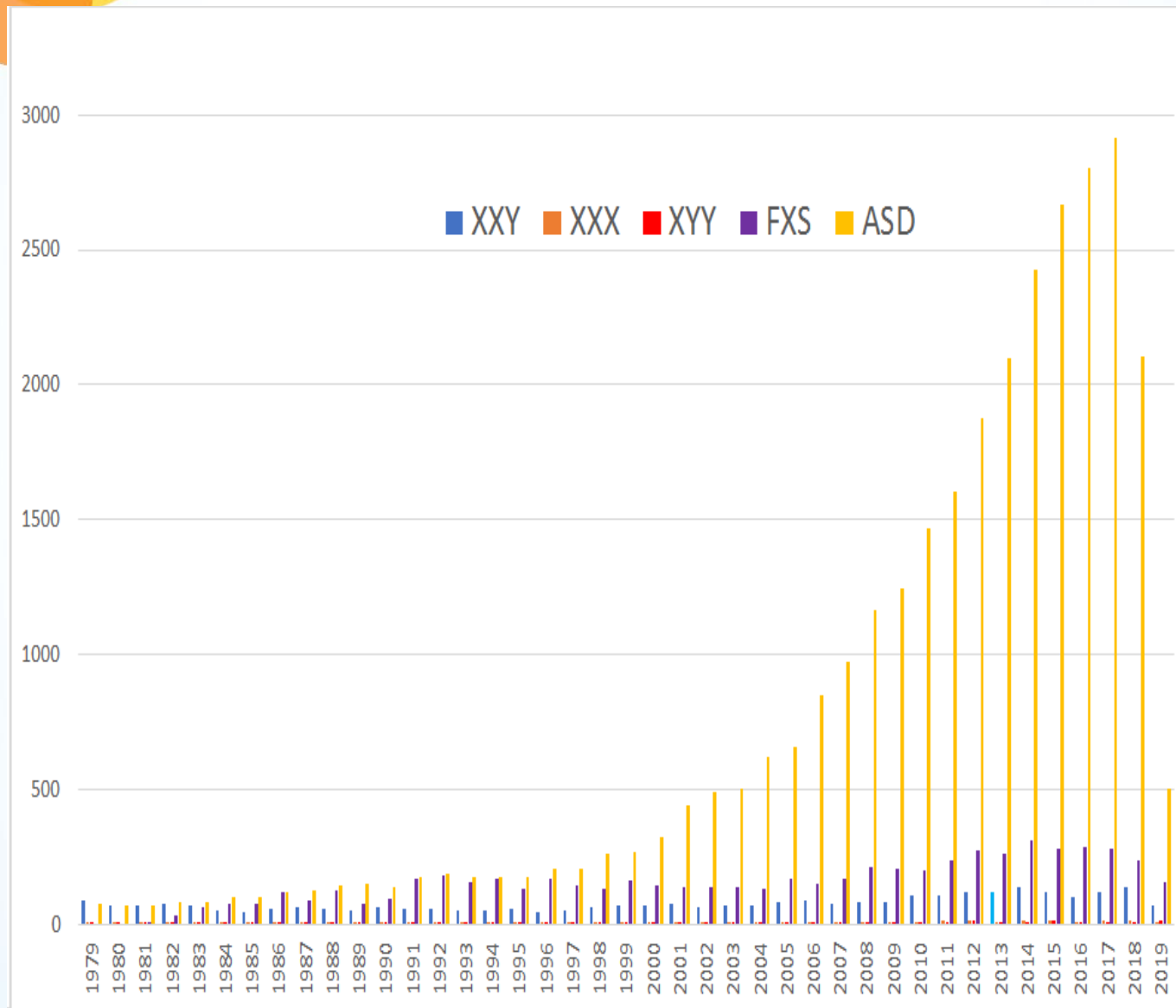


Fragile X Syndrome 6,261 Publications*



[*Source: US National Library of Medicine National Institutes of Health](#)





What can just one research paper do?

Review | **Open Access**

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

Received: 9 June 2009 | Accepted: 11 May 2010 | Published: 11 May 2010

Abstract

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

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Metrics

Article accesses: 132391

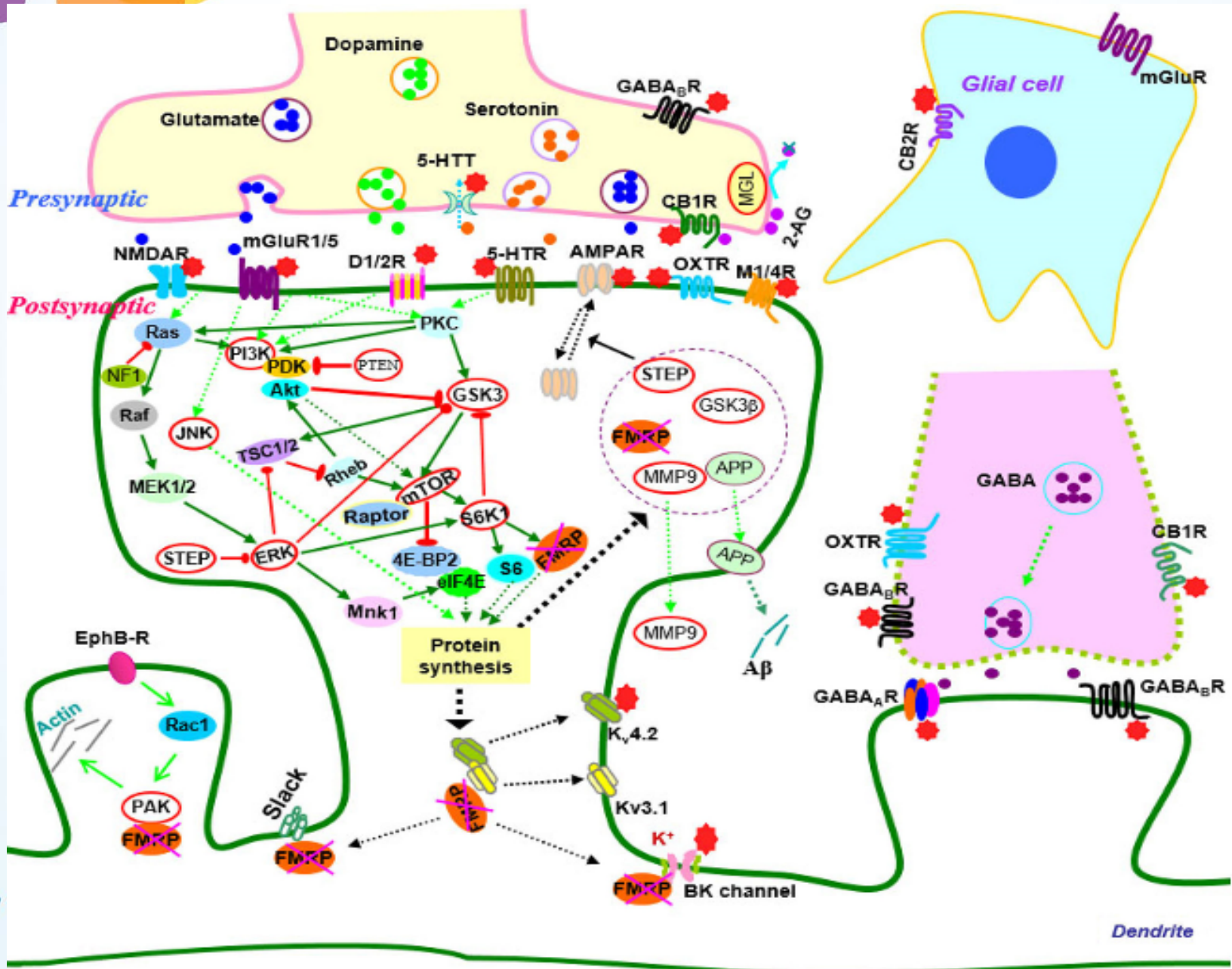
Citations: 92
[more information](#)

Altmetric Attention Score: 7

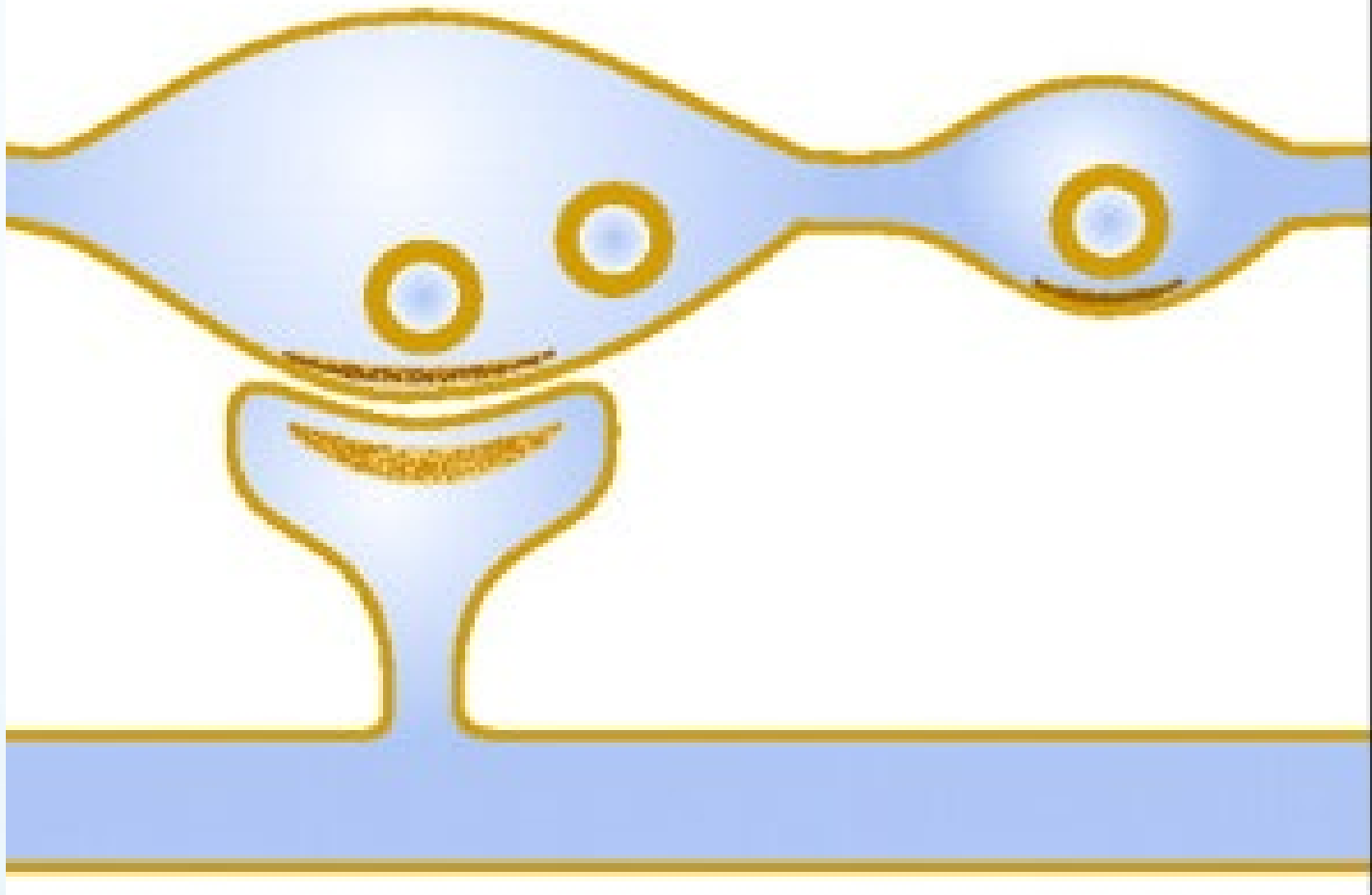


36 participants led to international impact





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



Research



