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Demographic Data of Patients seen in a Medical Genetics Clinic for Autism Spectrum Disorders (ASD) as part of an ongoing study on “Genetic Variations in ASD”.

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Demographic data of patients seen in a medical genetics clinic for Autism Spectrum Disorders (ASD) as part of an ongoing study on “Genetic Variations in ASD”.

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**EAST TENNESSEE STATE
UNIVERSITY**

Background

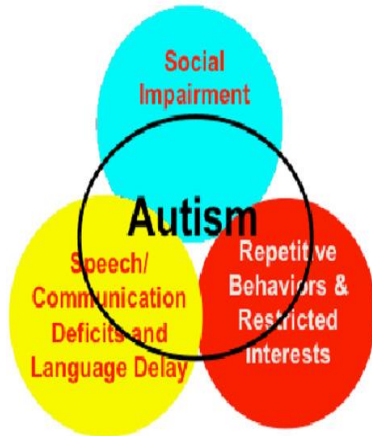
- Autism Spectrum Disease (ASD)
 - a lifelong developmental disability marked by deficits in social communication and social interaction, and restricted, repetitive patterns of behavior, interests, or activities.¹
 - Include Autism, Asperger's syndrome, and Pervasive developmental disorder (PDD).^{1,2}



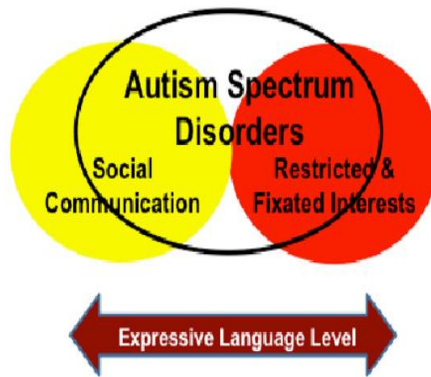
Background

Classification

A. **DSM IV:**
Pervasive Developmental Disorders:
Autism



B. **DSM5:**
Autism Spectrum Disorders



Associated features



<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3446247/>

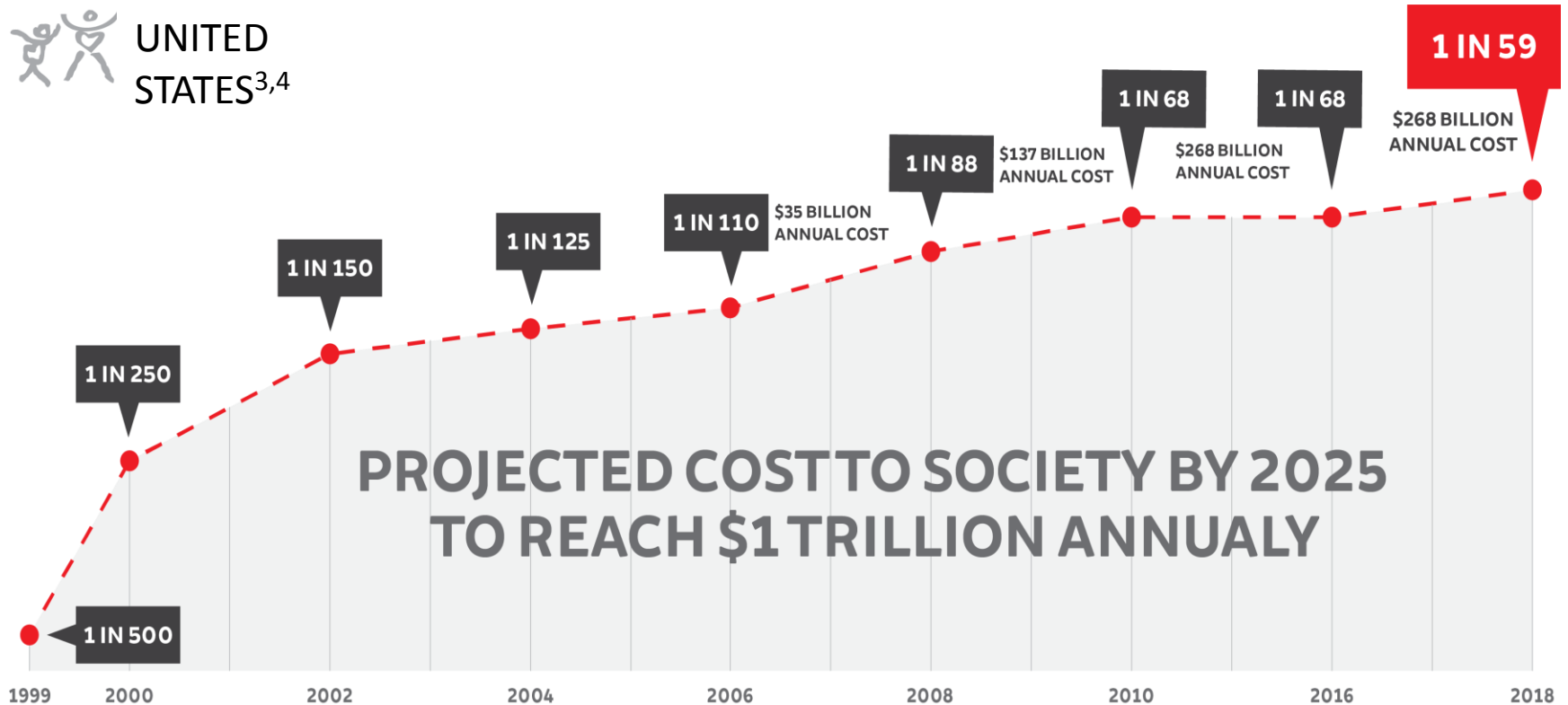
<https://blogs.biomedcentral.com/on-biology/2018/04/03/diagnosis-treatment-autism-real-story/>



Background



UNITED STATES^{3,4}

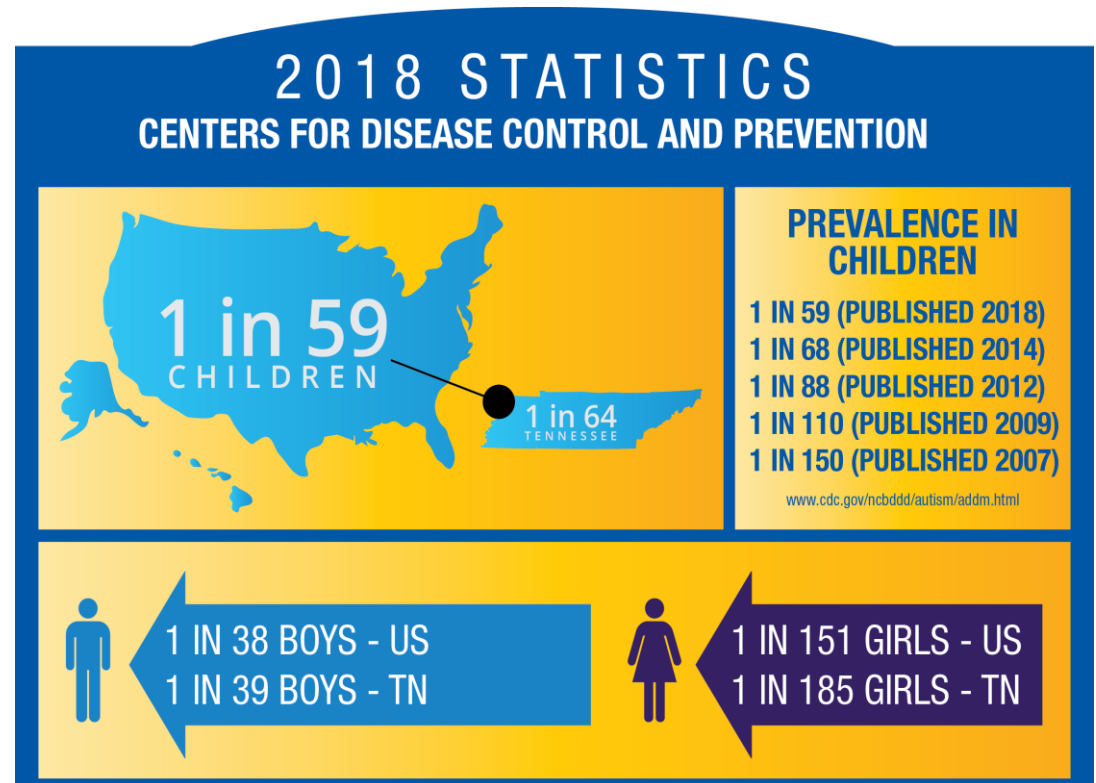


<https://taconow.org/autism-statistics/>



Background

- ASD has been found to be about 4-5 times more prevalent in males, compared to females. ¹⁻⁴
- Increase in the prevalence of congenital abnormalities in patients with ASD.⁵



<https://vkc.mc.vanderbilt.edu/vkc/triad/autismdata/>



Purpose

- Influenced by the DSM V classification and consequent projections.
- An ongoing multi-year retrospective chart review of the patients seen at the Genetics Clinic for suspected or confirmed cases of ASD.
- With the preliminary data, we identified the demographic characteristics of patients and examined how they compare with documented studies.



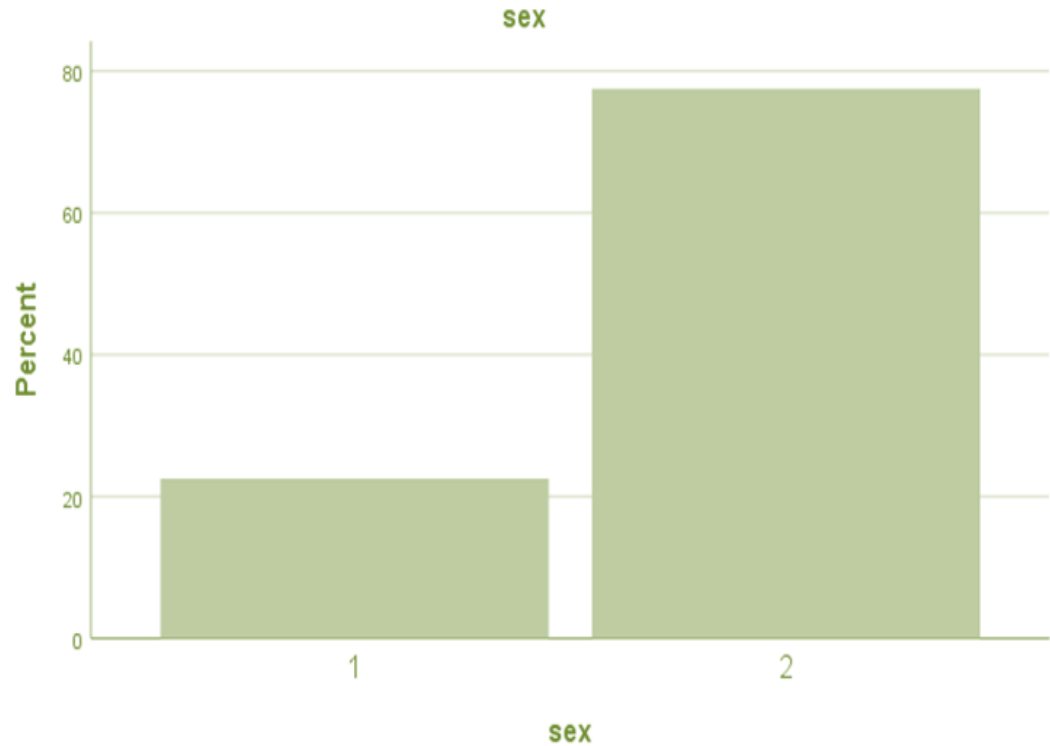
Methodology

- An ongoing multi-year, retrospective chart review.
- We extracted data from the Electronic health record (Allscripts) for 80 patients (n=80)
 - Gender,
 - Term status at birth,
 - Twin status,
 - Presence of congenital abnormalities,
 - State of residence.
- Data extraction tool-REDCap
- Data analysis by the Statistical Package for Social Sciences (SPSS).



Results

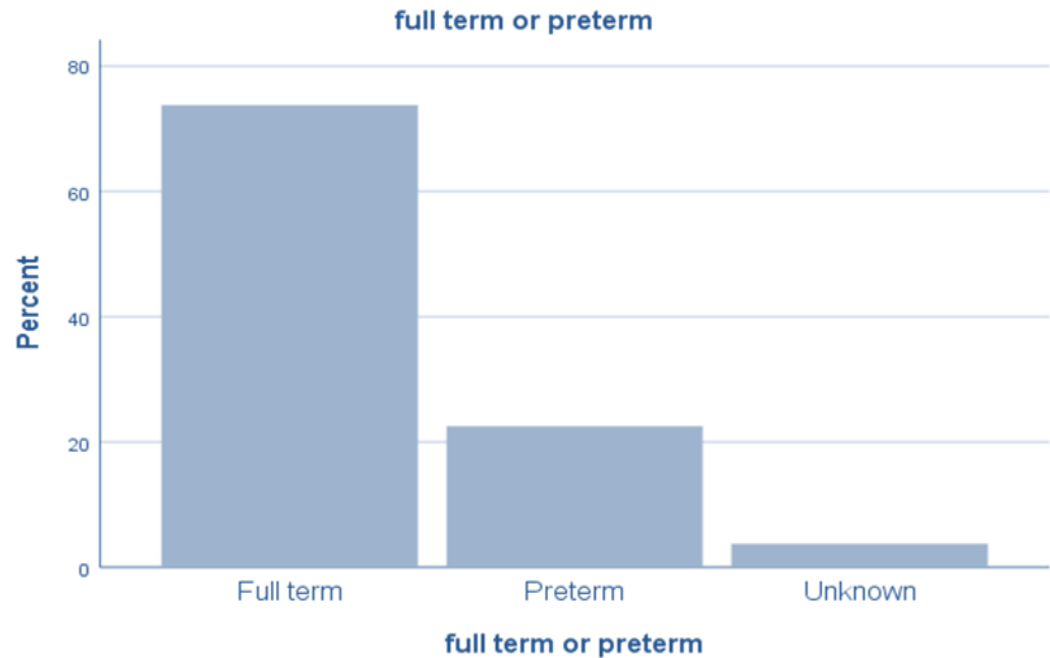
- Gender/sex
- 1= Female
 - 2= Male



Results

➤ Term status

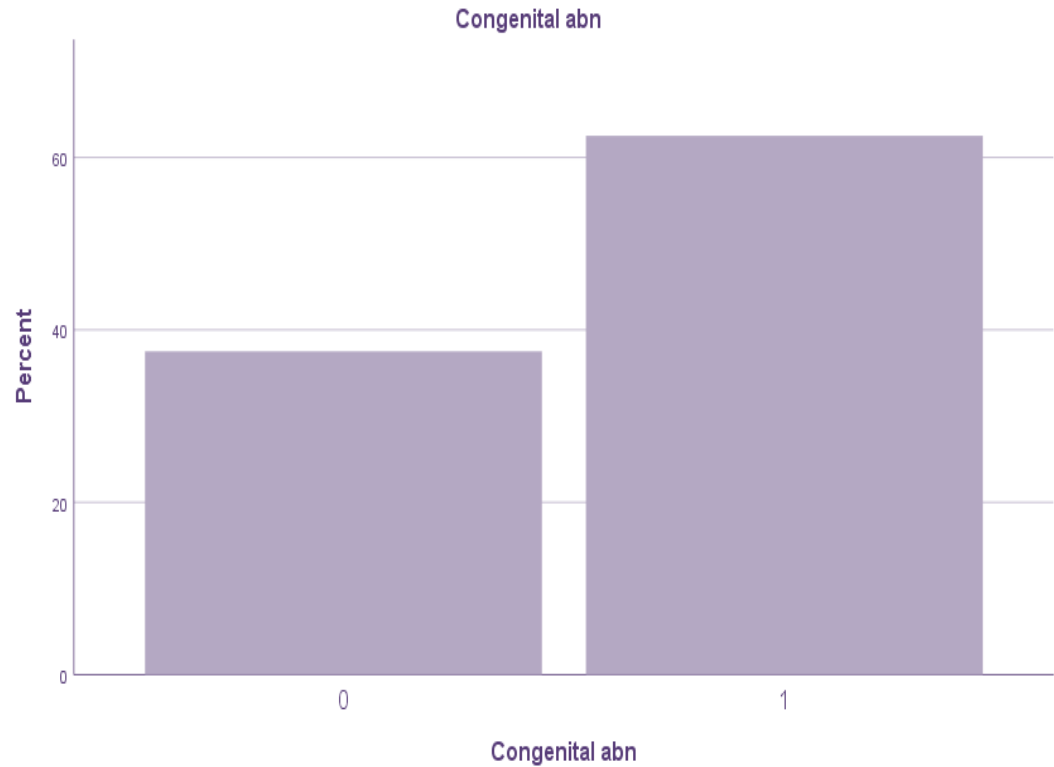
- Term: ≥ 37 weeks
- Preterm: < 37 weeks



Results

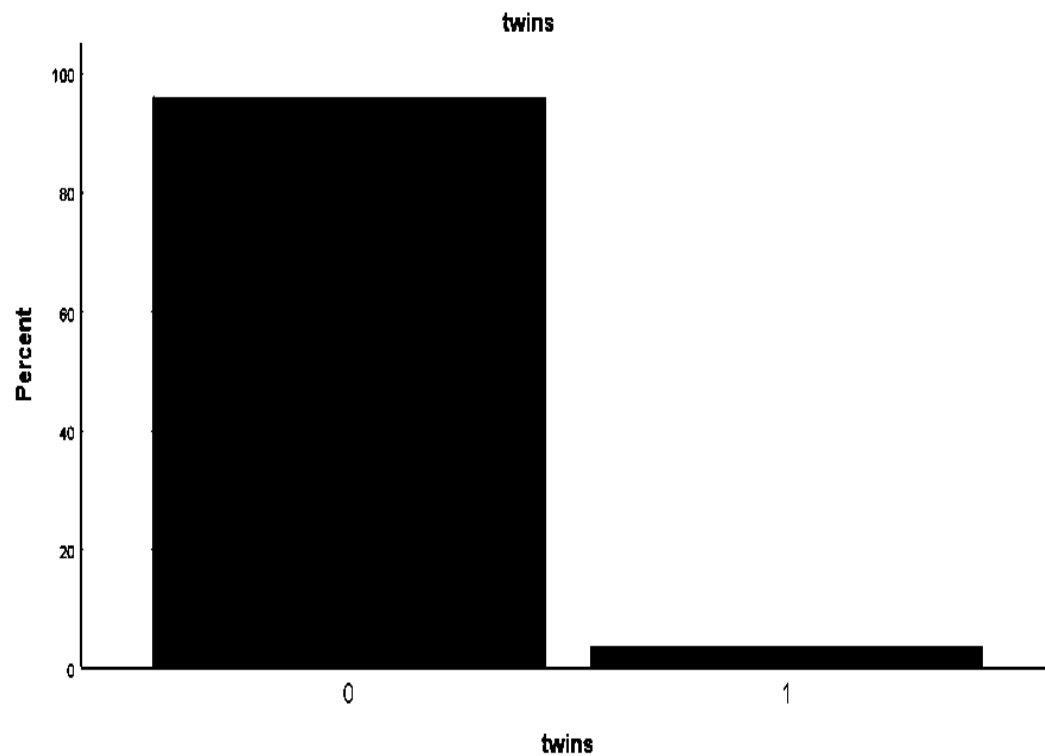
➤ Presence of congenital anomaly

- 0= No
- 1= Yes



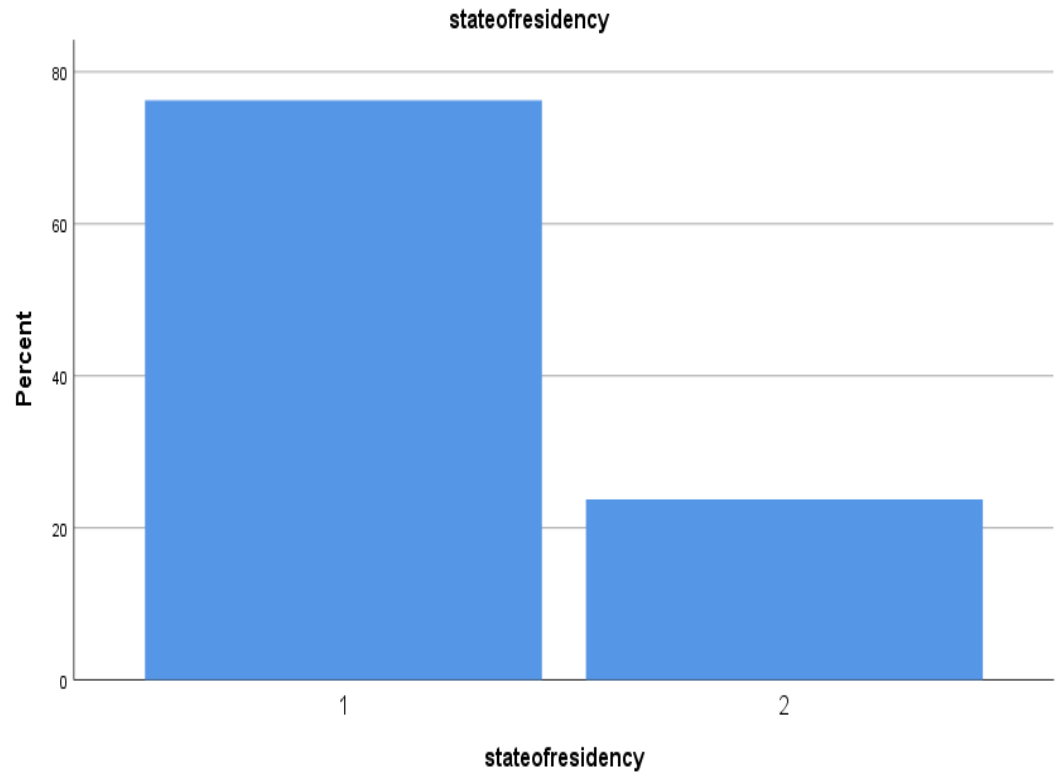
Results

- Twin status
- 0= Not a twin
 - 1= Twin



Results

- State of residency
- 1= Tennessee
 - 2= Virginia



Limitations

- Small sample size
- Limited demographic data
- Absence of baseline data as a basis of comparison



Conclusion

- Increased proportion of males compared to females.
- Increased proportion of the population with congenital abnormalities compared to those without such abnormality.
- These findings agree with documented demographic data from previous studies on ASD.



Implications

- Need to ensure more awareness on ASD.
- Funding for more research on ASD.
- Establishment of more genetic centers.
- Encourage early diagnosis:
 - Prompt management
 - Improve quality of life



Future directions

- We would examine the
 - Rate of diagnosis of ASD in the clinic,
 - Type of mutation and the genes involved
 - Trends for specific gene, and/or specific mutation



Acknowledgement

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- Rebecca Roberts
- Kathryn Duvall



References

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5. TIMONEN-SOIVIO L, VANHALA R, MALM H, et al. The association between congenital anomalies and autism spectrum disorders in a Finnish national birth cohort. *Dev Med Child Neurol*. 2015;57(1):75. doi:10.1111/DMCN.12581



THANK YOU

