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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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

- Social Impairment
- Repetitive/Communication Deficits and Language Delay

B. DSM5:
Autism Spectrum Disorders

- Social Communication
- Restricted & Fixed Interests

Associated features

AUTISTIC SPECTRUM DISORDER

- Autism
- ADHD
- Specific Learning Difficulties
- Anxiety
- Tourette's
- ODD
- OCD
- Sensory Integration Disorder
- Gifted

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

UNIVERSITY STATES

https://tacanow.org/autism-statistics/
Background

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

https://vkmc.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: \( \geq 37 \) weeks
  • Preterm: < 37 weeks
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
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References


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