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# Tuberous Sclerosis Complex

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<https://www.youtube.com/watch?v=RJnbaOeXKds>

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# Learning Objectives:

- Description of Tuberous Sclerosis Complex (TSC)
  - Relationship to Autism
  - Treatment Implications
  - (Mis)Understanding of TSC
  - Future Directions
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# What is TSC?

- Multisystem genetic disorder characterized by benign (noncancerous) tumor growth in multiple parts of the body
- Resulting from mutations in one of two genes, TSC1 or TSC2
- Tumors form primarily in the brain, kidney, eyes, skin, lungs, and heart



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# Prevalence

- Prevalence ranges from about 1 in 6,000 to 1 in 10,000 births
  - But many cases remain undetected
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# What Does TSC Look Like?

- Signs and symptoms of TSC vary from person to person
  - Symptoms can include:
    - Skin abnormalities
    - Seizures/Epilepsy
    - Kidney/heart/lung/eye problems
    - Cognitive/Behavioral difficulties
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# Other Disabilities Related to TSC

- Developmental delays
  - Intellectual disability
  - Learning disability
  - Autism
  - ADHD
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# Relationship to Autism

- ASD exists more frequently in individuals with TSC than in general population
- TSC is one of the leading genetic causes of autism; May be due to:
  - Nonspecific disruption of brain function due to tubers
  - Seizures
  - Disturbances in brain development in areas associated with ASD
  - Cognitive impairment



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# Relationship to Epilepsy

- Epilepsy may be the most challenging, and prevalent, manifestation of TSC
  - Occurs in about 70-80% of patients
  - In most cases, the region in which the seizure originates is the location of a cortical tuber
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# Cognitive and Behavioral Implications

- The cognitive and behavioral problems associated with TSC tend to be the greatest concerns to caregivers
  - A wide range of behavioral difficulties may be seen
    - Aggression
    - Temper tantrums
    - Restlessness
    - Impulsivity
    - Concentration difficulties
    - Self injury
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# Interventions

- Early diagnosis and early intervention for TSC can help reduce related developmental delays
  - Important to accurately diagnose ASD in children with TSC, so they can receive educational and medical support as early as possible
  - Complex needs of individuals with TSC need to be assessed and managed by multiple disciplines
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# (Mis)Understanding of TSC

- Many TSC cases go undiagnosed for years due to lack of awareness and because symptoms can be mild (not as easily detected)
  - Unlike other genetic disorders that result in distinct physical appearances and specific symptoms, TSC results in symptoms that can vary from person to person, so no two individuals with TSC look exactly alike
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# Future Directions

- Need better detection of TSC
- Future research might include:
  - Better definition of neurobiological basis of autism in TSC
  - Efficacy of interventions for autism in children with TSC
  - Relationship of treatment response to cognitive abilities and frequency of seizures
- Longitudinal research is needed on:
  - Mechanisms underlying cognitive and behavioral deficits
  - Interventions
  - Biomarkers of cognitive and behavioral disorders
  - Lifespan progression and outcomes of individuals with TSC, developmental trajectories



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# To Conclude

- Many people with TSC lead healthy lives as independent individuals
  - TSC has a wide clinical spectrum and many patients show minimal symptoms  
or may have no neurologic disabilities
    - Most people with TSC live normal life spans
  - Monitoring throughout the lifetime by medical professionals is important
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