Autism and public health
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Autism is a spectrum. Some suggest calling it autisms.

Prevalence

- “... the best estimate of current prevalence of ASDs in Europe and North America is approximately 6 per 1000”
  - Johnson et al., 2007
- Prevalence by type:
  - Autism Disorder - 2.2 per 1000
  - Asperger Disorder - 1.0 per 1000
  - PDD-NOS - 3.3 per 1000
  - Fombonne et al., 2006
**The basics**

- ASD symptoms (social responsiveness, communication, need for sameness) vary
- Caused by interacting factors – genetic changes, interacting genes, epigenetic factors (influences on genes), and environmental stressors - that disrupt these faculties
- Common biochemical, structural or developmental pathways may be impacted at different places and by different agents, to cause ASD

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**Early Warning Signs**

- A developmental / diagnostic evaluation is indicated if:
  - There is any loss of language or social skills at any age
  - The child does not
    - babble or coo by 12 months of age
    - gesture (point, wave, grasp, etc.) by 12 months
    - say single words by 16 months of age
    - say two-word phrases on his or her own (rather than just repeating what someone says to him or her) by 24 months of age

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**Screening and Early Identification**

- AAP Recommended Surveillance and Screening Algorithm
  - Evaluate risk factors:
    - Is there a sibling with autism spectrum disorders?
    - Are parents concerned?
    - Are other caregivers concerned?
    - Are you, as the child's physician, concerned?
  - If at least two risk factors present and child is at least 18 months old, administer ASD specific screening tool.

  Johnson et al., 2007
### Important Genes

- Gene mapping in consanguineous families or families with multiple persons with ASD
- Across >12 linkage studies, most consistent evidence for 7q22-q32, but not in largest study
- Genome wide association studies
- Autism Genome Project Consortium

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### Where we are today

- Several of the observed deleted genes are regulated by neuronal activity
- Prenatal development is guided by intrinsic gene expression patterns
- Brain continues to develop after birth, and experience and environmental input impact subsequent development
- Synapses (connections between neurons) mature as function of experience-dependent neuronal activity and gene-expression changes that go with it
- Dysregulation of synaptic development – a predominant theme in autism research
- It is unlikely to be "one thing" most of the time.
<table>
<thead>
<tr>
<th>Do I do an autism genetic test?</th>
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<tbody>
<tr>
<td>- Consider referral for syndromes and targeted testing</td>
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<tr>
<td>- Chips probably not ready for clinical utility, but how will you decide?</td>
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<tr>
<td>- Where do you put resources – early identification, services, research, other?</td>
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