Genetic Conclusions

- The notion of finding a simple genetic cause to help define ASD will not likely occur.
- There are likely over 1,000 genes that have various roles in ASD which replicates the huge clinical variability of the ASDs.
- Genetic information is confirming the notion that there are “autisms” rather than an “autism”, multiple distinct disorders analogous to “cancers”.
- Nevertheless it may be possible to define subtypes of ASDs based on the patterns of certain genetically determined molecular pathways that might be targets for specific treatment.

Layers of the ASD Onion

- Layer of DNA
- Layer of the Brain
- Layer of the Mind
- Layer of the Community

BRAIN GROWTH

Three Phases of Growth Pathology in Autism

Courchesne, Webb, Schumann, 2010
The brain in ASD

- Neurobiological disorder...a disorder based in the brain.
- Autopsy, neuropsych testing, imaging, EEG, MEG, TMS, etc all show differential activity in the brain in ASD.
- 3 current theories:
  - Connectivity hypothesis
  - Social information processing hypothesis
  - Social motivation hypothesis
Interconnectivity Hypothesis

- Due to poor long range connectivity, simple, local processing is intact while complex, distributed information processing is impaired.
- The nature of the information processed is relevant only insofar as it requires distributed brain function.
- Because social interaction tends to be complex, these theories suggest that it is particularly vulnerable to disruption due to underconnectivity.

Support for Interconnectivity

- Imaging study results vary: showing overconnectivity, underconnectivity, and typical connectivity in ASD.
- Behavioral correlates are proposed: improved visual-spatial, splinter skills, etc.
- Movement artifact in MRI actually causes increased close connectivity and decreased long range connectivity: blow to connectivity theories.
- Connectivity unlikely to be universal.

Imitation Impairments in Autism

- Imitation impairments long noted in autism
- Some consider imitation deficit a core impairment of ASD
- Only two of twenty well designed studies fail to find impairments
- Observed very early in life, prior to two years of age

- Symbolic meaning hypothesis (Baron-Cohen, 1988; Rogers et al, 1996)
- Executive functioning hypothesis (Ozonoff, Pennington & Rogers, 1991; Rogers et al, 1996)
- Poor social motivation to attempt tasks (Trevarthen & Aitken, 2001)
- Dyspraxia or motor dysfunction (Jones & Pryor, 1985)
- Deficit in Self-Other mapping (Rogers & Pennington, 1991; Williams et al, 2001; Williams et al, 2004)
Imitation and Mirror Neurons

• The cellular substrate for imitation may be the mirror neuron system
• The mirror neuron system has been studied most in non-human primates
• These neurons activate during execution of an action as well as the observation of an action
• Mirror neurons appear to be part of the “social brain”

Mirror Neurons

• Mirror neurons may provide the mechanism through which we have the ability to understand other’s emotions and actions via an “internal representation” without need to reflect.
• Thus this may be the neurologic mechanism to underlie imitation, empathy, theory of mind, metaphor and evolution of language
• Mirror neuron function can be studied through EEG, fMRI, MRI and TMS measurements - Sara Webb
Conclusions about the brain in ASD

- Brain growth is abnormal early on in ASD maybe due to abnormal connectivity of critical brain regions needed for social communication
- Something about the function of the social brain is amiss in ASD
- Imitation is a mechanism via which we can understand other's behavior. Understanding how imitation works in the human brain (mirror neurons) may lead to treatments that improve social understanding

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Theory of Mind
By 2 months old a child is “programed” to look at eyes.

Figure 5.4
A schematic summary of the “Sally–Anne Test” of understanding false belief. (C = child; E = experimenter.) Reproduced from Baron-Cohen, Leslie, and Frith 1985.