



Autism

- ❑ Autism involves self-absorption
- ❑ Specific features of autism include
 - Impaired social relations with others
 - An inability to communicate
 - Literal interpretation of speech
 - Impaired imaginative ability
 - Stereotyped movements
- ❑ Incidence of autism is 4/10,000
 - Males are 4 times more likely to develop autism

- L'autismo è un disturbo dello sviluppo, riconosciuto come tale dal DSM
- La sua caratteristica principale è relativa ad un'immaturità dell'area socio-affettivo-emotiva
- Il bambino autistico evita lo sguardo , ogni forma di contatto fisico e di interazione sociale



RAIN MAN





- Generalmente non usa il linguaggio per comunicare
- Tende ad isolarsi
- E' molto ripetitivo nei comportamenti e disturbato dai cambiamenti e dalle novità che non tollera facilmente
- Si evidenziano le stereotipie
- Parla in terza persona e non in prima persona
- Ripete meccanicamente frasi o la parte finale di una frase

- E' evidente un uso improprio di giochi come pure la tendenza ad usare un giocattolo in modo ripetitivo e senza variarne l'uso
- Assente il gioco simbolico, di finzione
- Intanto che cresce si rivela incapace di cogliere comportamenti scherzosi e battute di scherzo
- Si rivela incapace di prevedere i comportamenti altrui (teoria della mente)

Caso 6
Anna
4 anni

Scarsa contatto oculare. Non considera gli altri e li urta come se non esistessero. Talvolta, è troppo attaccata alla madre. Fornisce risposte incoerenti agli stimoli verbali. Preferisce il gioco solitario. Dimostra ecolalia e inversione dei pronomi. È incapace di iniziare o sostenere una conversazione. Allinea i giocattoli. Spegne e accende la luce. Oscilla ripetutamente la mano. Si copre le orecchie quando è arrabbiata. Sviluppa episodi acuti di agitazione e aggressività.

Caso 7
Ronald
8 anni

Ignora le persone. Urta gli altri bambini, come se non esistessero. Dimostra scarsa contatto oculare. "Non sa come rivolgersi ai coetanei e farsi accettare da loro". Cita argomenti irrilevanti e fuori contesto. Non intraprende alcun tipo di gioco immaginativo. Ha utilizzato parole singole fino all'età di 3 anni. È ecolalico. Allinea i giocattoli. Gli interessi e le attività sono notevolmente ristretti. È iperattivo.

Insorgenza all'età di 3 anni. Ammiccamenti e tic facciali; si tocca ripetutivamente il viso, spostando indietro i capelli; emette rumori simili a grugniti.

Padre: ammiccamenti. Zio: tic della testa. Nonno: smorfie con la bocca, ammiccamenti e episodi deliranti. Prozia: smorfie facciali e delirio di essere una musicista di fama mondiale. Prozio: afferma di controllare il clima con una macchina. Zio materno: difficoltà sociali da adulto; isolamento sociale durante gli anni della scuola. Zio: irrequietezza, tamburellamento continuo con le dita. Nonno: difficoltà sociali.

Insorgenza all'età di 6 anni. Ammiccamenti e scuotimenti della testa e delle spalle; mette in atto in modo ripetitivo comportamenti come battere le mani, tamburellare con le dita, schiarirsi la gola e emettere rumori schioccanti.

Padre: scuotimenti della testa e delle spalle; ammiccamenti. Non lavora. Zio paterno: schiarimento continuo della gola, scuotimenti della testa. Non lavora. Nonno paterno: bizzarro, impulsivo, irrequieto.

Tic motori

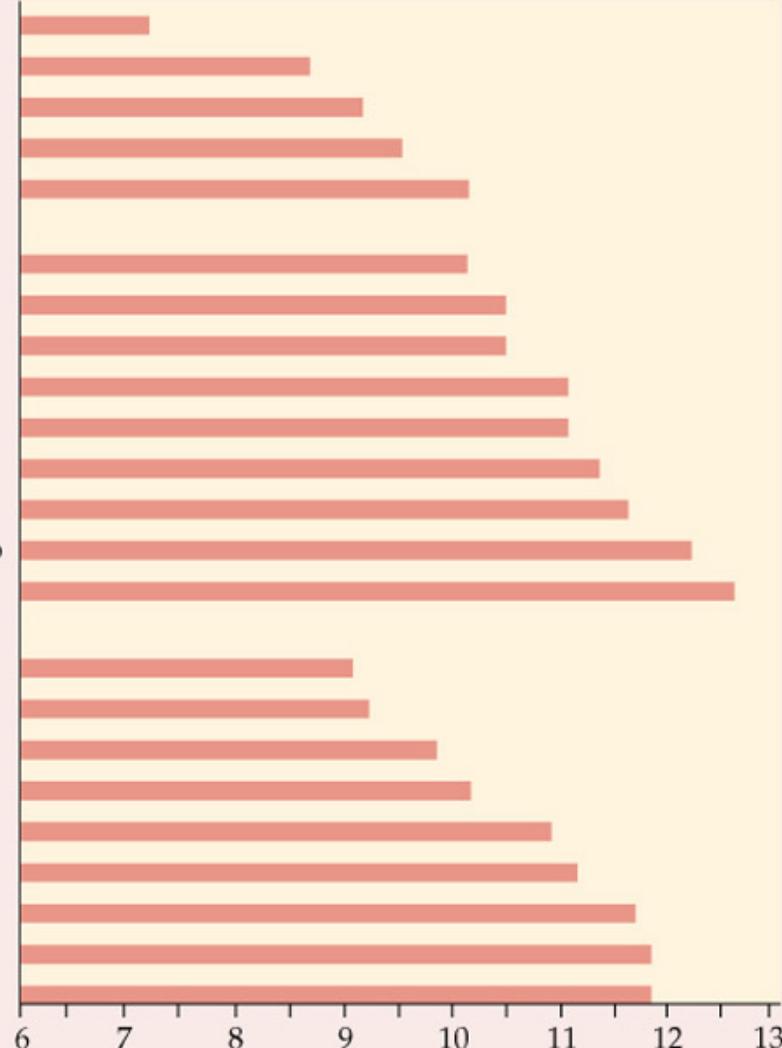
- Occhi, faccia, testa
- Spalla, collo
- Braccia, mani
- Tronco
- gambe

Tic vocali

- Rumori modesti
- Rumori elevati
- Balbattare
- Ripetizione
- Coprolalia
- Sillaba
- Blocchi
- Parole al di fuori del contesto
- Ecolalia

Azioni compulsive

- Sbattere la testa
- Baciare
- Tccare gli oggetti
- Scalciare
- Battere
- Toccare se stessi o altri
- Mordersi
- Toccare gli organi genitali
- Mimare gli altri

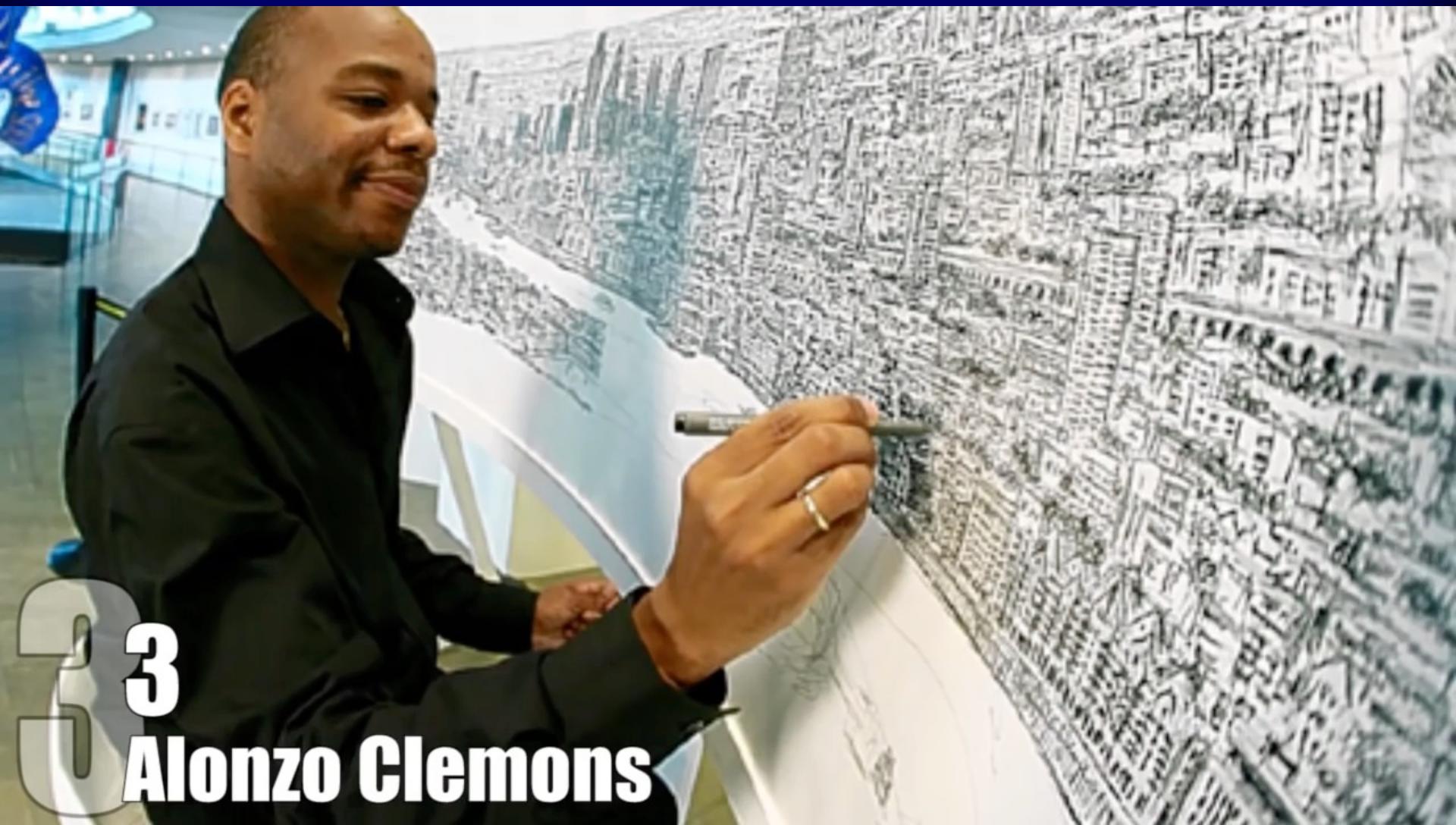


Età media all'esordio (anni)

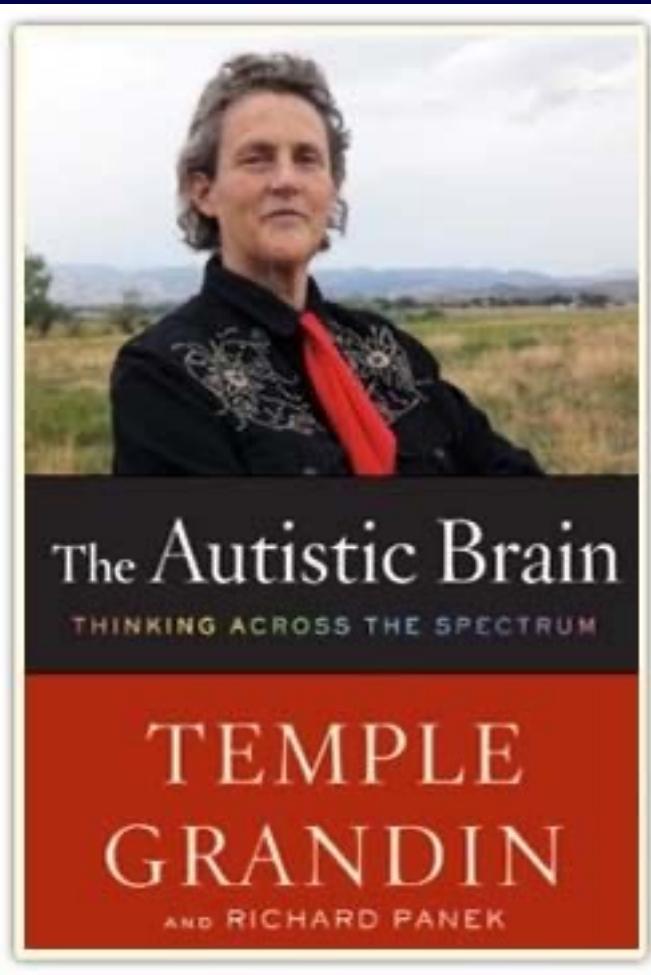
Savant skills

- ten times more common in people with autism than in others with mental handicap
- occurring in approximately one in ten individuals with autism

Savant



33
Alonzo Clemons



Temple Grandin



Biological Bases of Autism

□ Heritability of autism

- MZ twins exhibit a **70%** concordance rate for autism, while that of DZ twins is comparable to that of normal siblings
- 2-3% of the siblings of autistic children are themselves autistic

□ Autism is associated with neurological disorders:

- Phenylketonuria (PKU)
- Tourette's syndrome
- Fragile X syndrome (mental retardation)

□ Factors that impair development lead to autism:

- Rubella, hydrocephalus
- Drugs such as thalidomide (exposure during prenatal days 20-24 impairs development of the brainstem)

- Possible Causes:
- Biological:
 - Research and mental health professionals are convinced autism is caused by biological factors
 - Between 2 and 3 percent of siblings of people with autism are themselves autistic
 - There is a 70 percent concordance rate for monozygotic twins

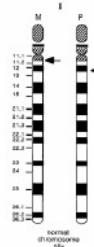
- Possible Causes:
- Brain pathology:
 - Heritable aspect of autism suggests the disorder is a result of structural or biochemical abnormalities in the brain
 - Researchers have found evidence for structural abnormalities in the brains of autistics, but so far we cannot point to any single abnormality as the cause of the disorder

Ricerche

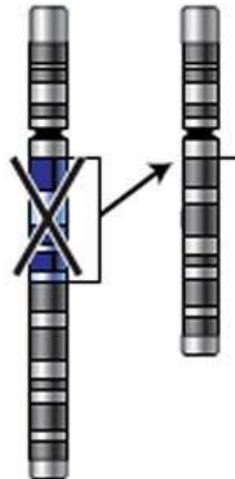
sui

fattori genetici

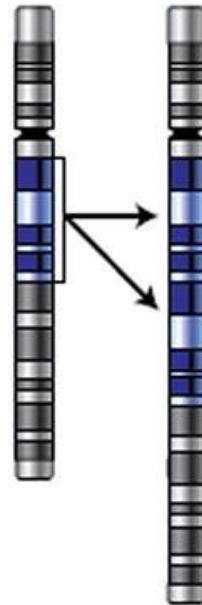
- Strong evidence of heritability
- Consistent 4:1 male-female ratio
- Likely genetic effects due to modifications of secondary or tertiary structures of DNA rather than primary changes in gene sequence (epigenetics)
- Current estimate is that between 5 and 100 loci involved



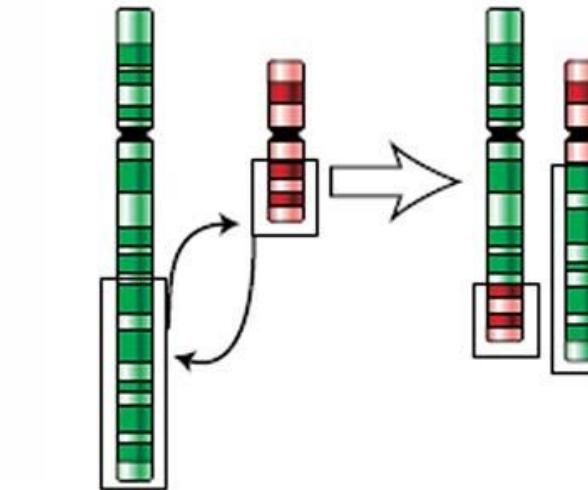
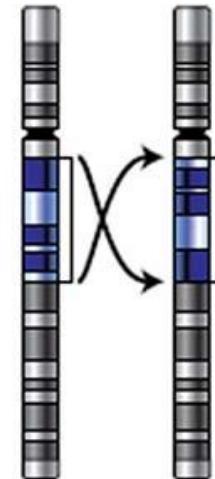
Deletion



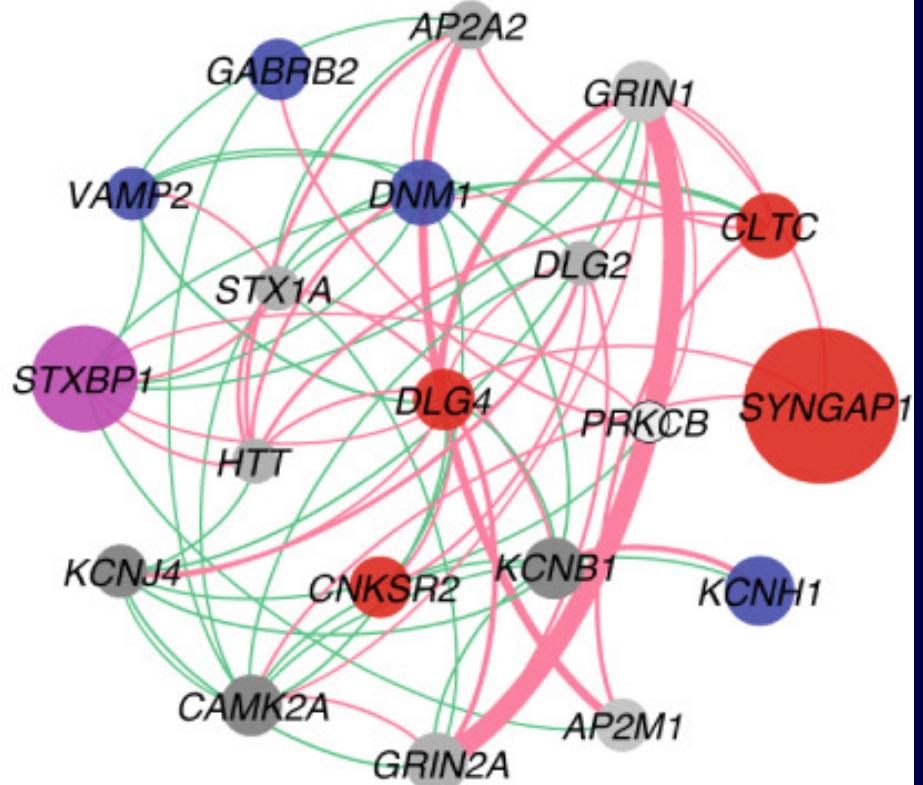
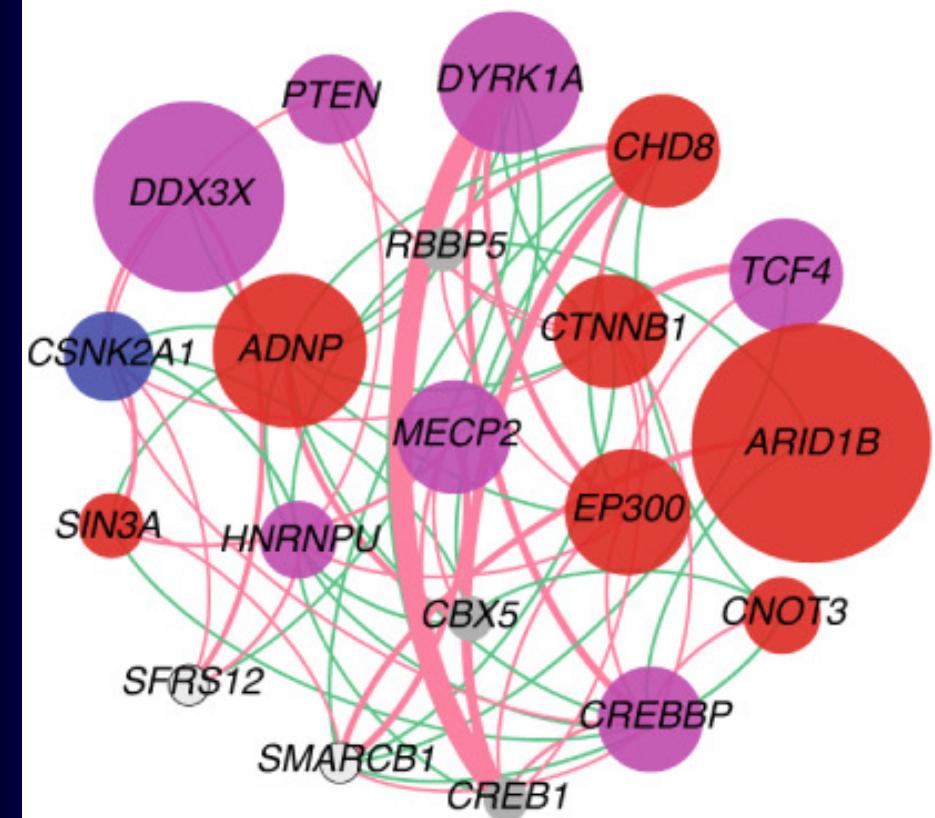
Duplication



Inversion



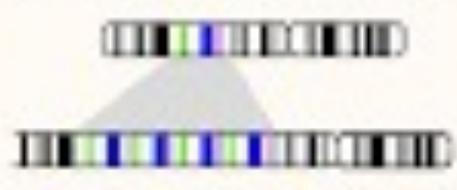
Reciprocal Translocation



Epigenetics



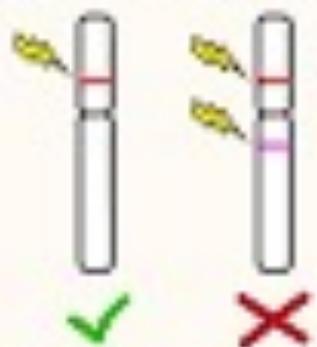
Copy Number Variation



Environment



Double-Hit Mutations

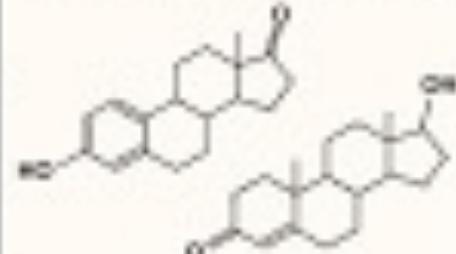


GENETIC SUSCEPTIBILITY

MOCO2, SHANK1, SHANK2, SHANK3, CHNAP9, SYNT1, SYNT2, C4DNL4T6, C4DNL82, KCNQ2, KCNQ5, KCNQ2, SYNUAPT, SUARSL3, USH2A, ALG13, ARXIN, ATRX, SCN2A, SMARDCA1...



Sex-Linked Modifiers



Autistic Spectrum Disorders

- Autism is one of five disorders coming under the umbrella of Pervasive Developmental Disorders (PDD)
 - Autistic Disorder
 - Asperger's Disorder
 - Childhood Disintegrative Disorder (CDD)
 - Rett's Disorder
 - PDD-Not Otherwise Specified (PDD-NOS)

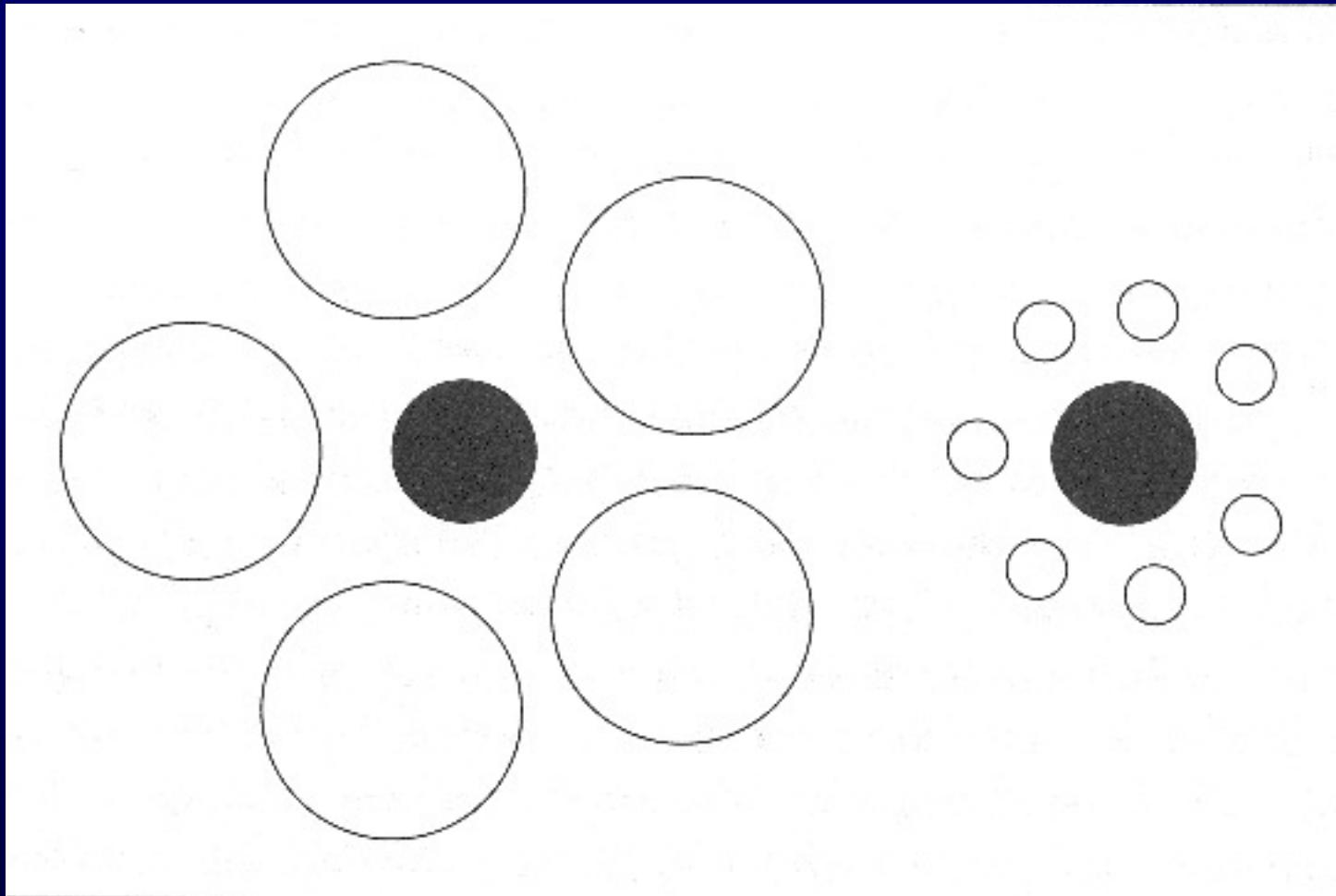
La Sindrome di Asperger spiegata in 8 min (dal Film "Adam")

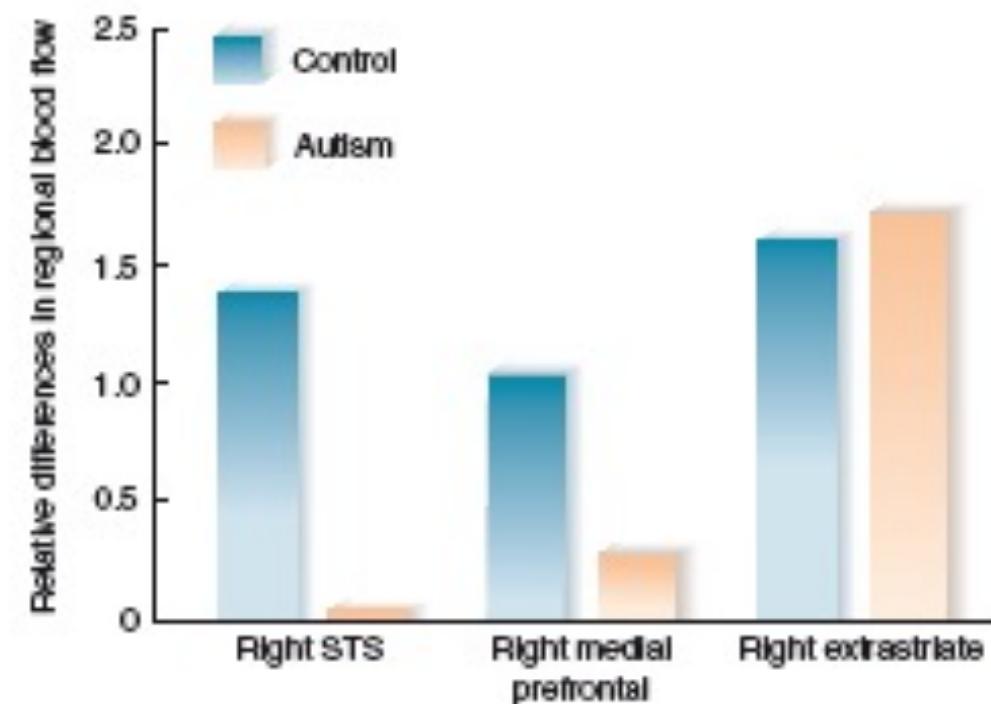
[https://www.youtube.co
m/watch?v=EXHaq5V6j
z8](https://www.youtube.com/watch?v=EXHaq5V6jz8)

Inadeguato sviluppo di una “teoria della mente”



L'illusione di Ebbinghaus





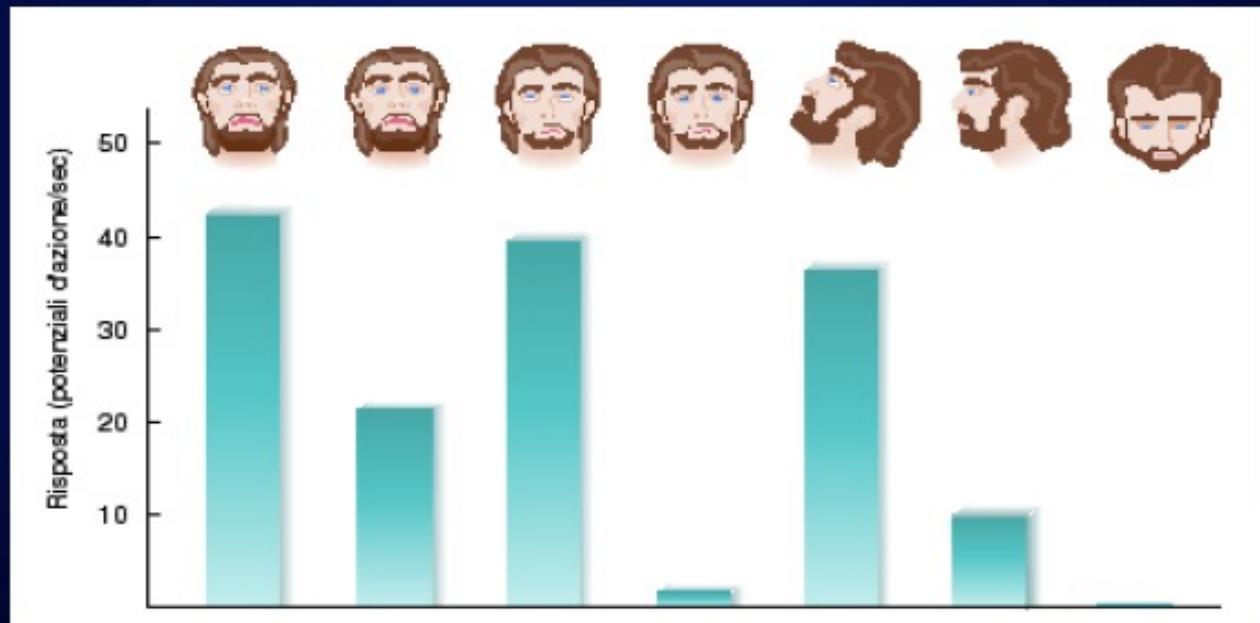
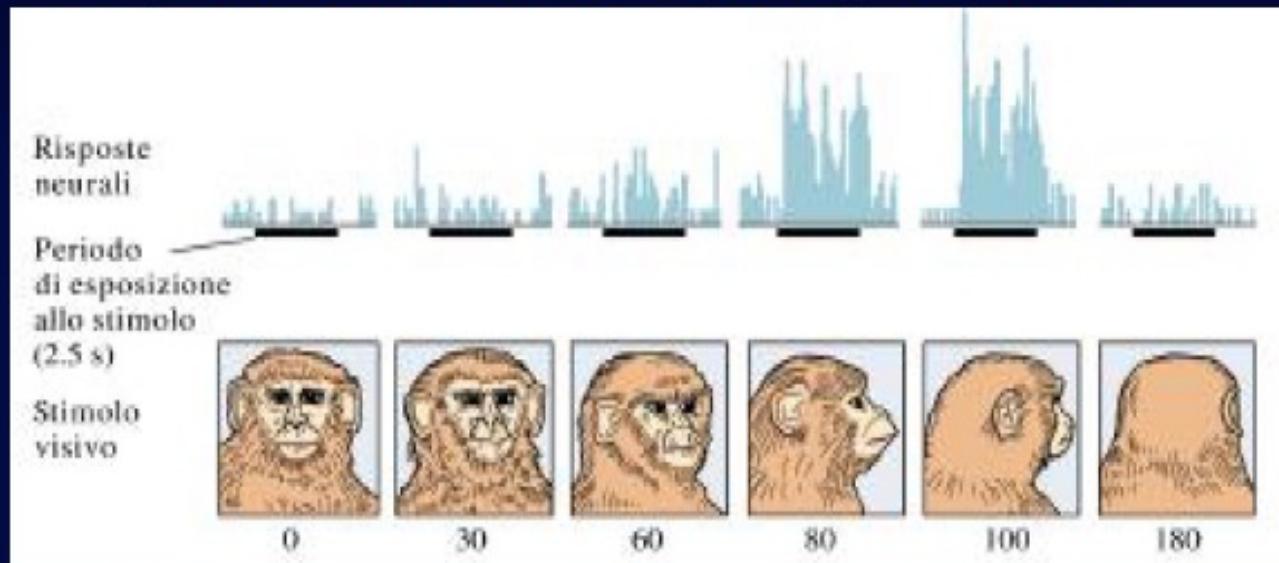
Regions with Significant Increase in Cerebral Blood Flow

FIGURE 17.5 Theory of Mind

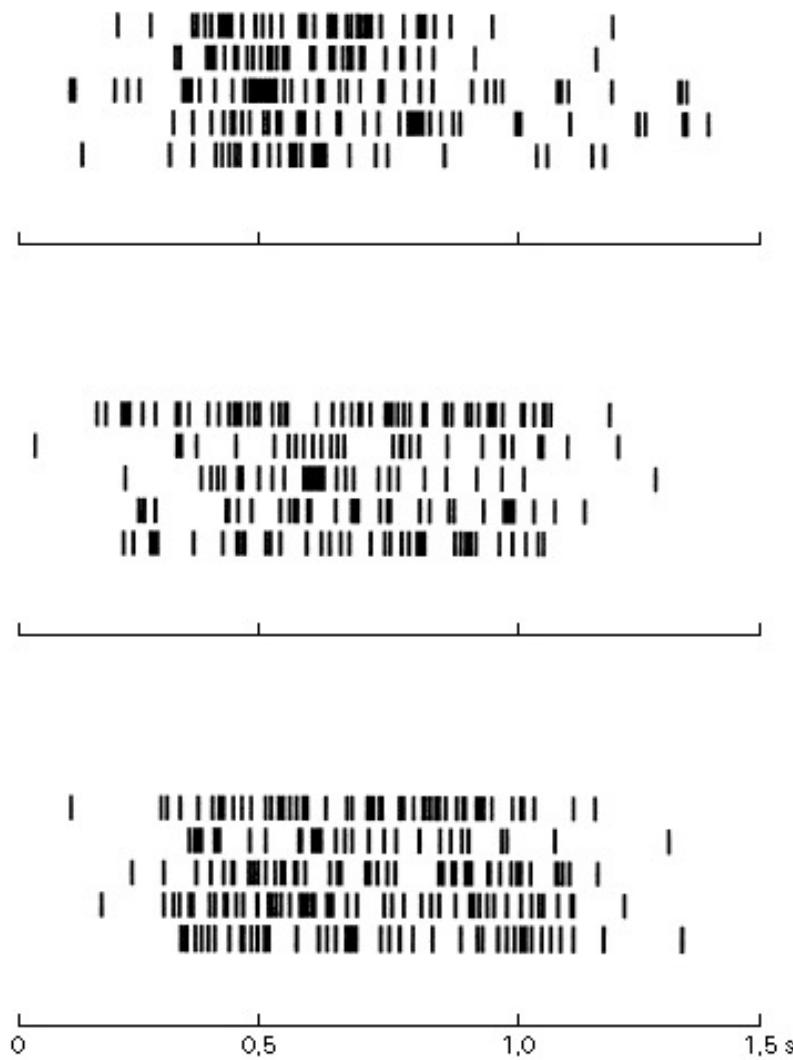
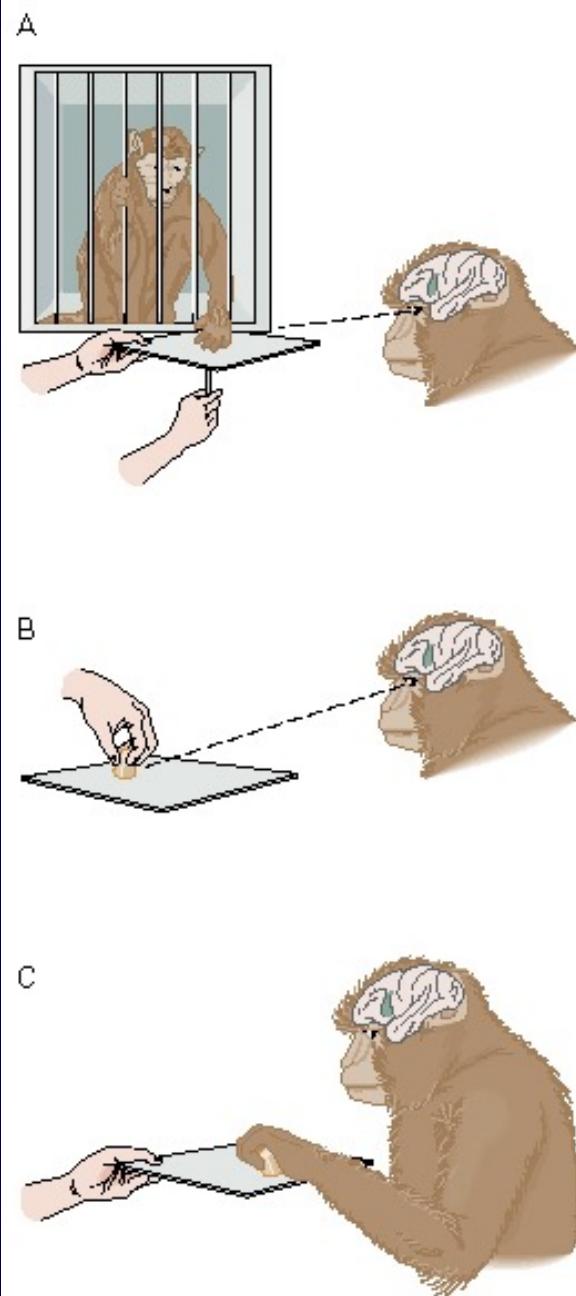
The graph shows relative activation of specific brain regions of autistic adults and normal control subjects viewing a "theory of mind" animation of two triangles moving interactively with implied intentions. STS = superior temporal sulcus.

(Based on data from Castelli et al., 2002.)

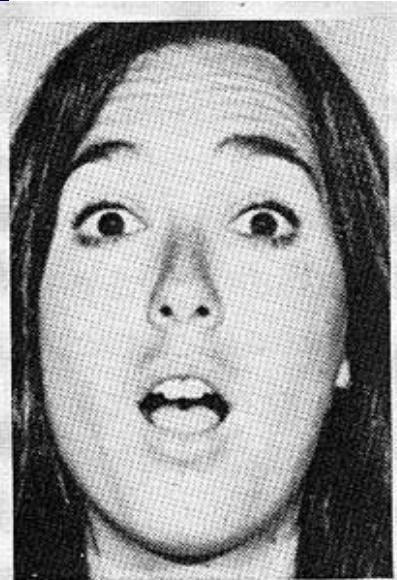
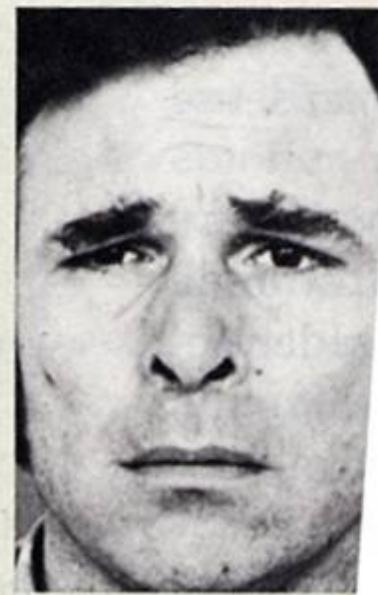
“Rispecchiarsi nell’altro” per capire l’altro



La scoperta dei neuroni *mirror*



Basic Emotions



Emotion and Facial EMG

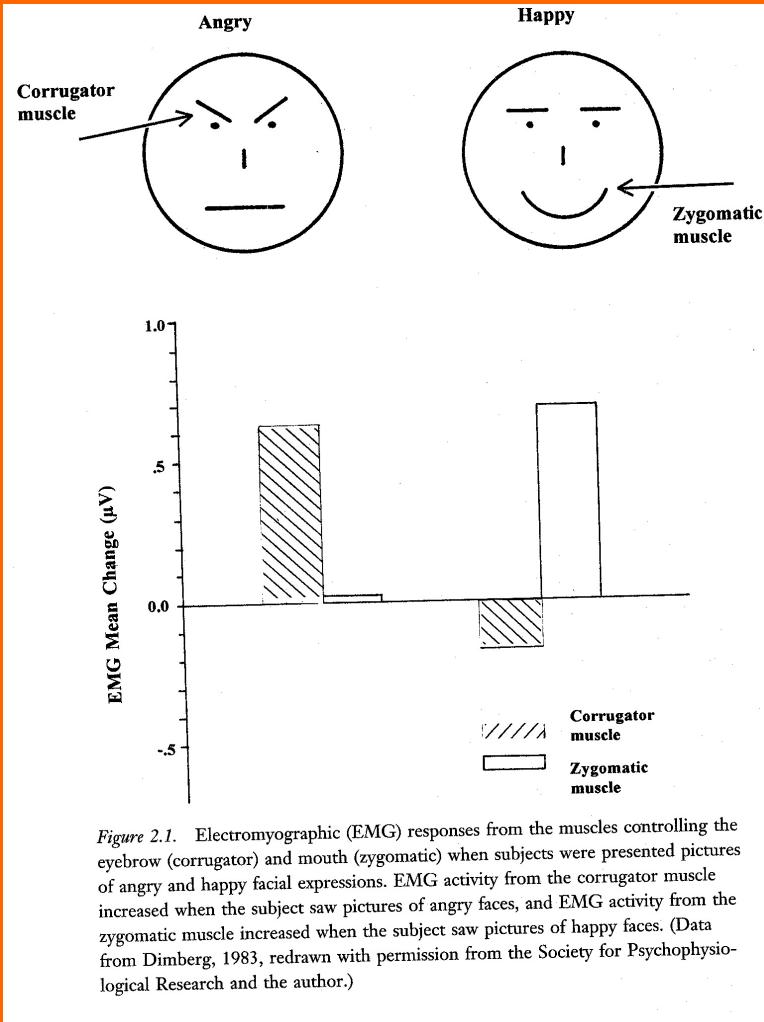
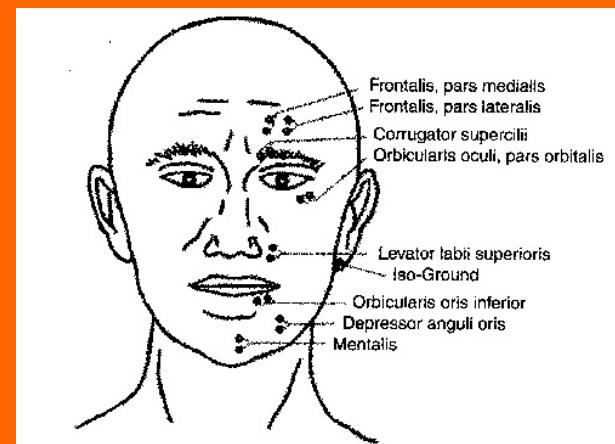


Figure 2.1. Electromyographic (EMG) responses from the muscles controlling the eyebrow (corrugator) and mouth (zygomatic) when subjects were presented pictures of angry and happy facial expressions. EMG activity from the corrugator muscle increased when the subject saw pictures of angry faces, and EMG activity from the zygomatic muscle increased when the subject saw pictures of happy faces. (Data from Dimberg, 1983, redrawn with permission from the Society for Psychophysiological Research and the author.)

- Corrugator
 - Negative affect
- Zygomatic
 - Positive affect



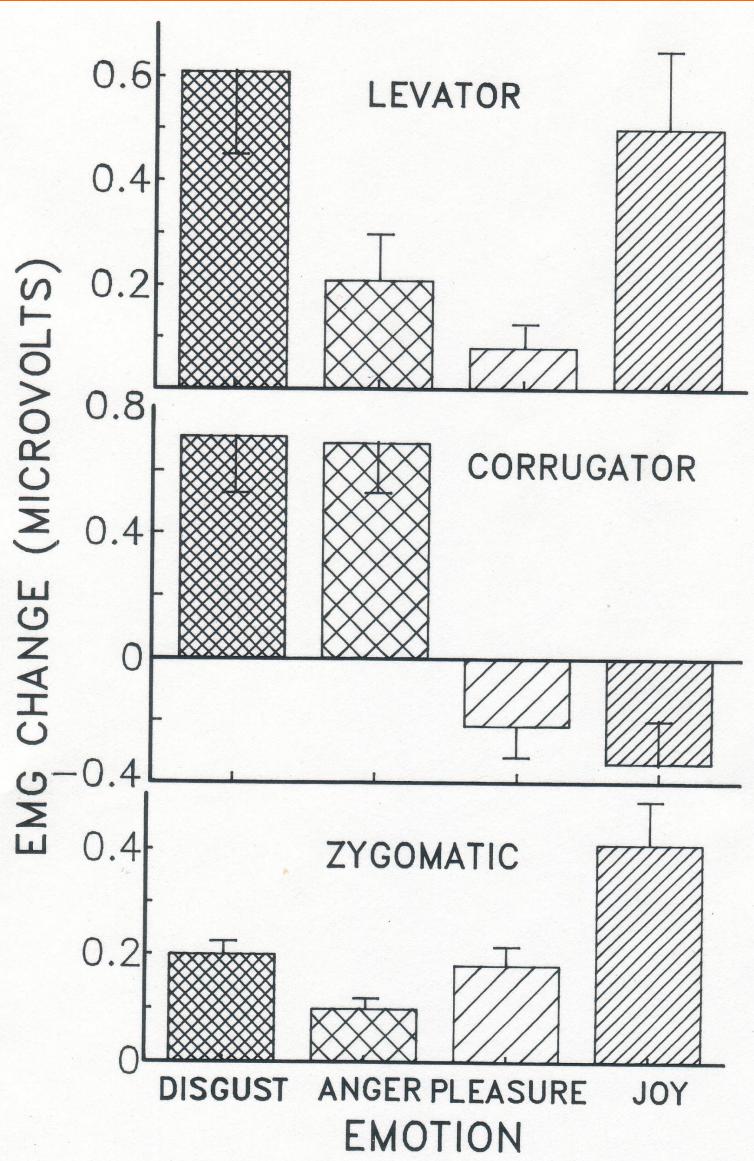
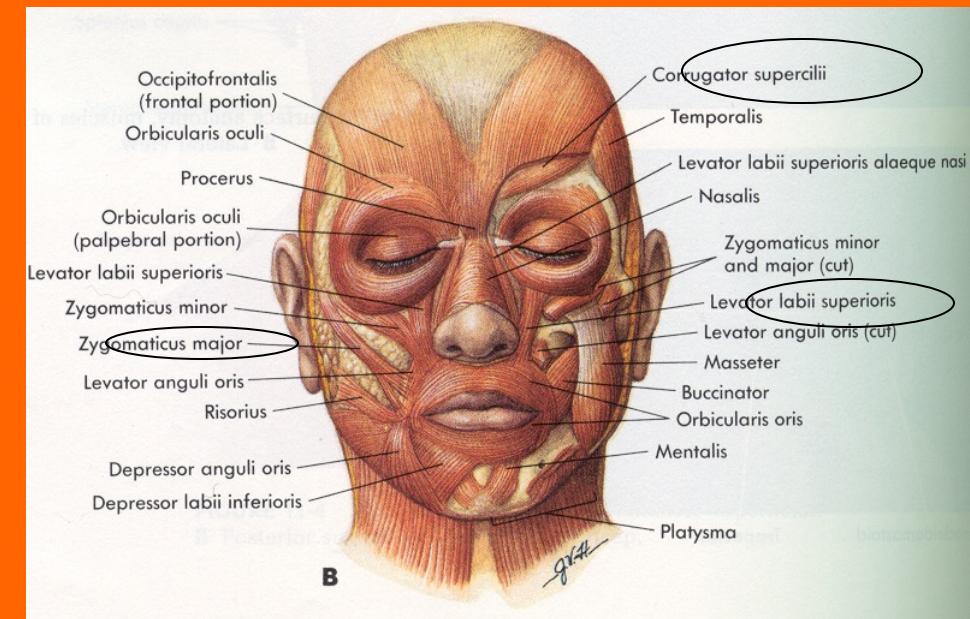
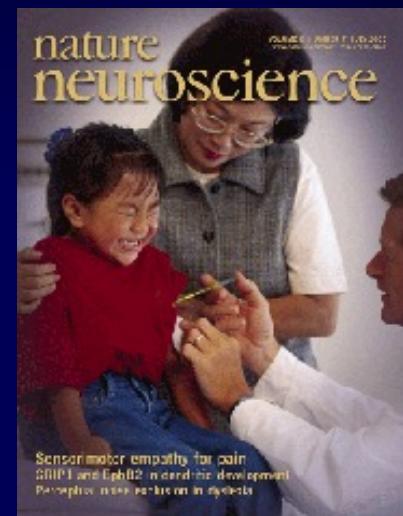
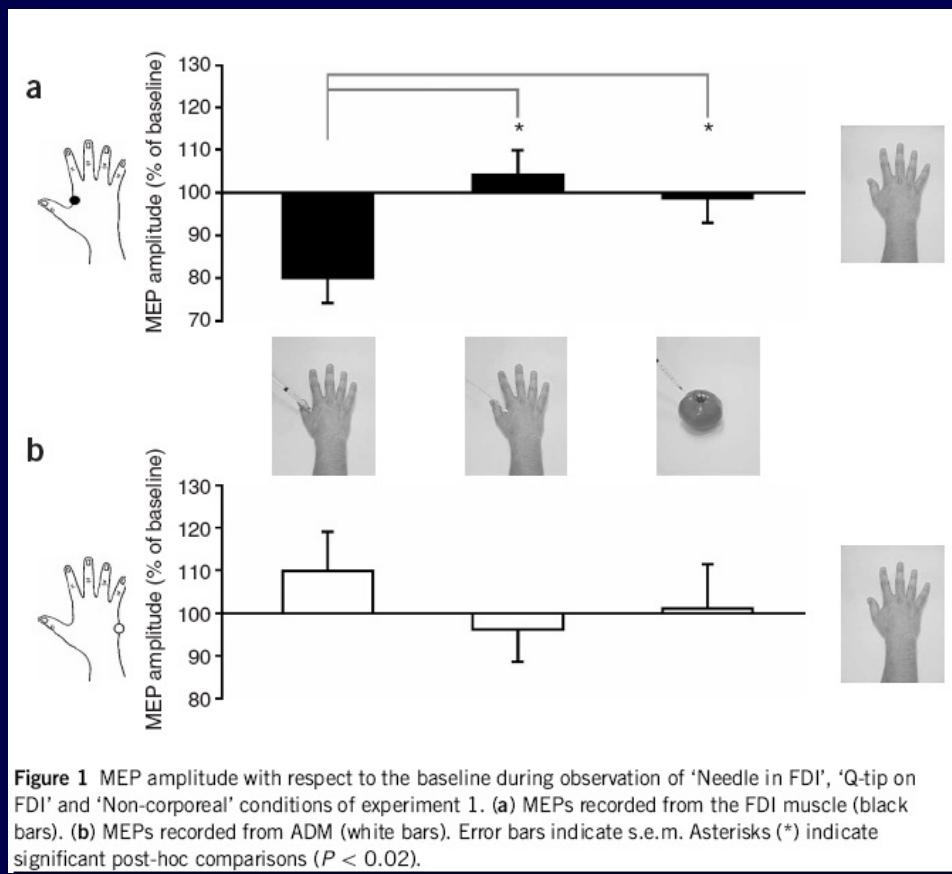
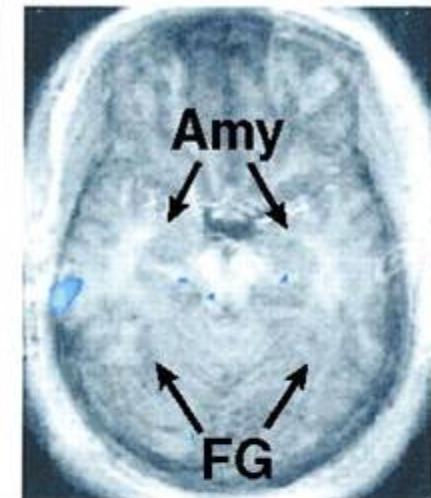
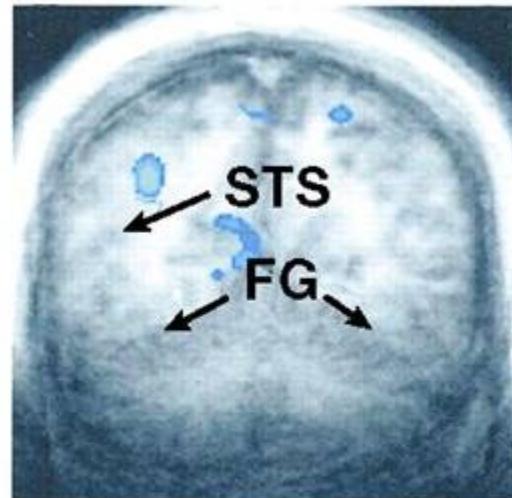
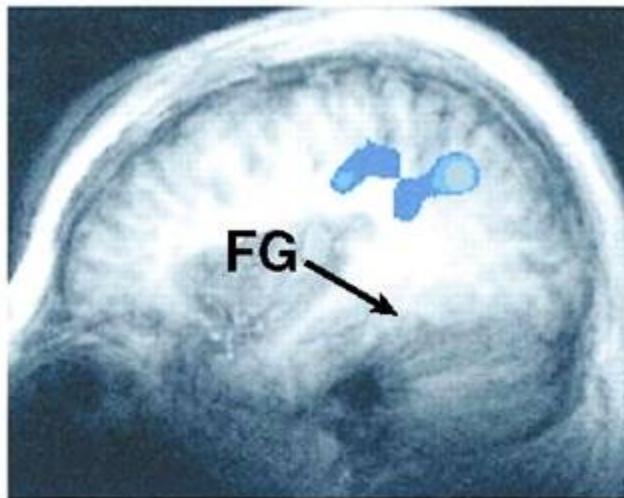


FIG. 8.7. EMG change from baseline and standard errors at the levator labii superioris/aleque nasi, corrugator, and zygomatic muscle regions as a function of emotional content of imagery. (Figure courtesy of Dr. Scott R. Vrana of Purdue University.)

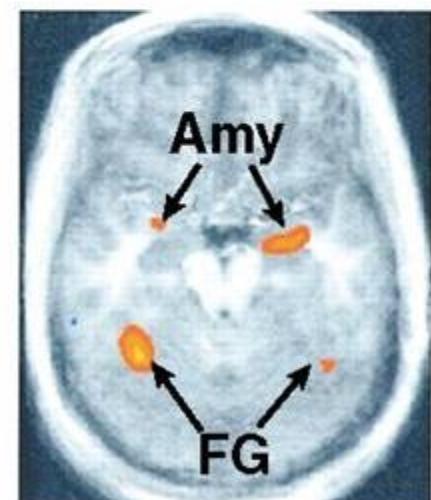
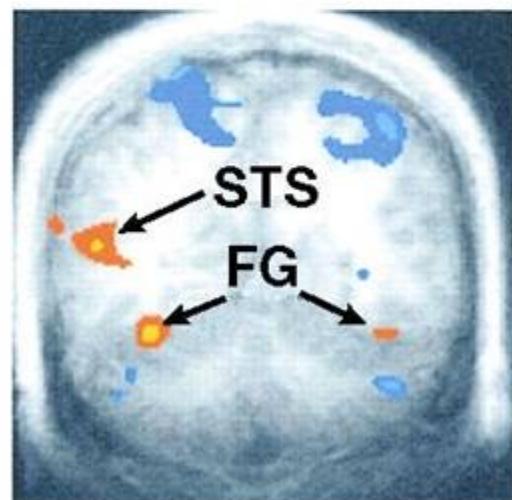
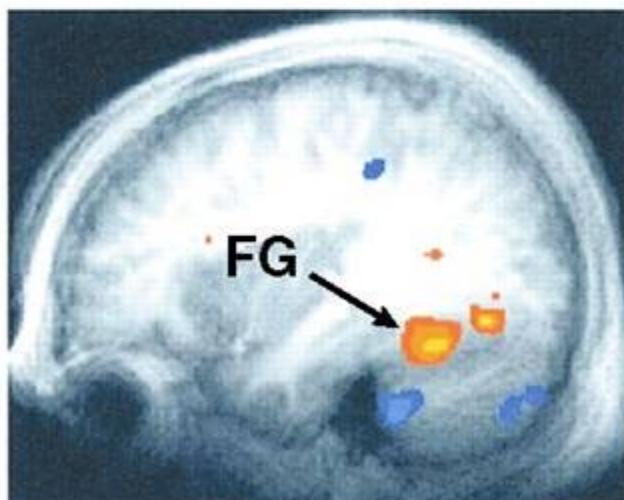




Autism



Normal



fusiform gyrus (FG)
superior temporal sulcus (STS)

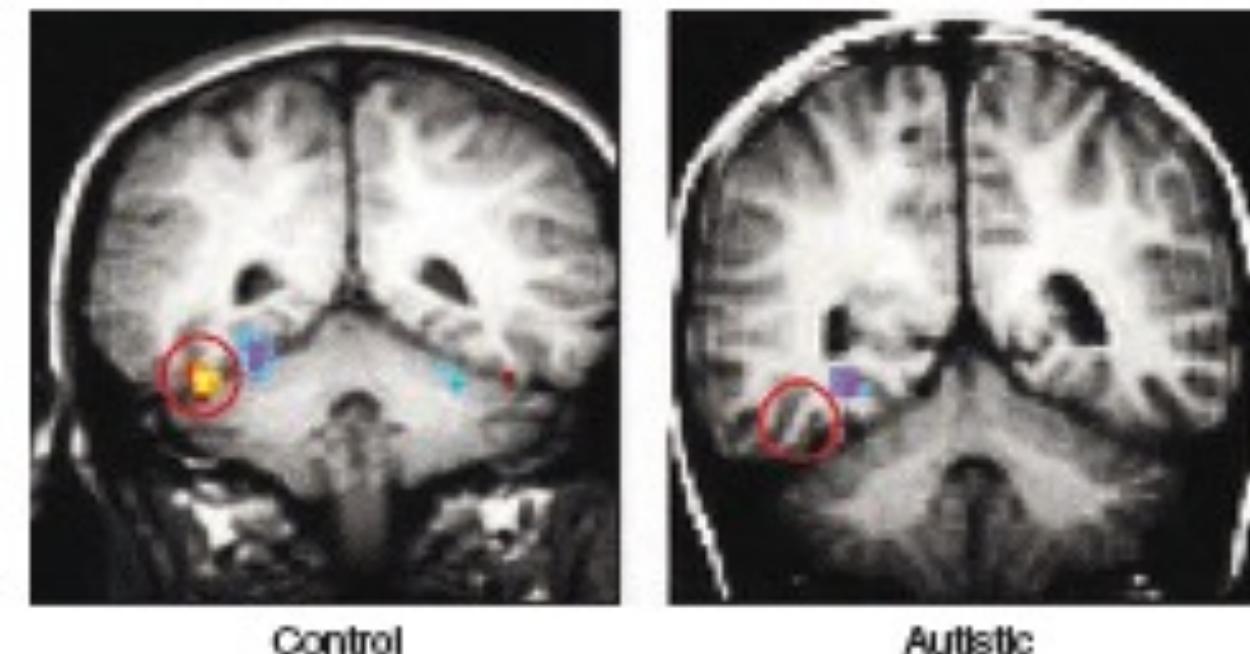


FIGURE 17.6 Fusiform Face Area and Autism

The scans show activation of the fusiform face area of control subjects but not of autistic subjects while looking at pictures of human faces.

(From Schultz, R. T. *International Journal of Developmental Neuroscience*, 2005, 23, 125–141. Reprinted with permission.)

fusiform gyrus (FG)

Pazienti autistici



A

B

C

D

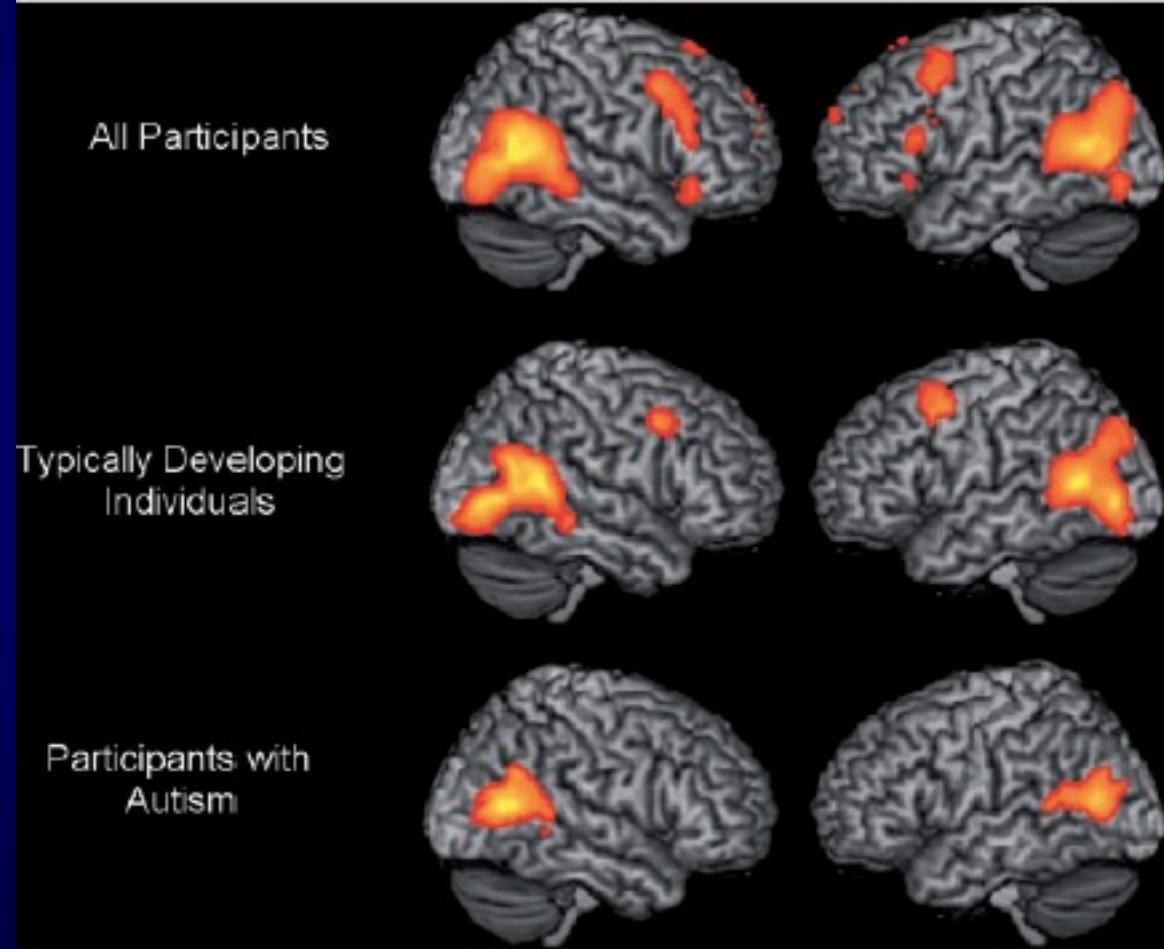
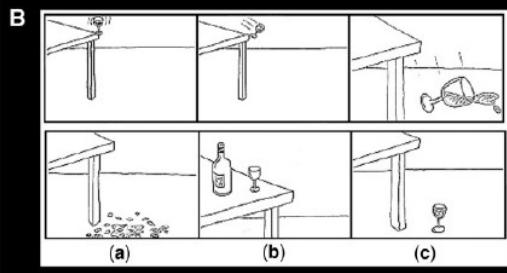
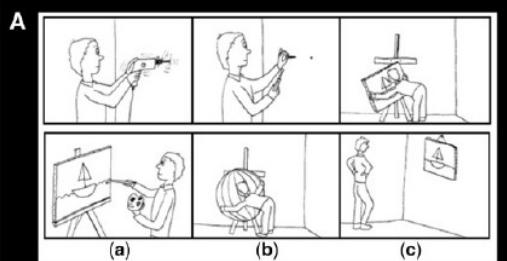
Faces on the left are data from two normal controls (A, B); faces on the right are data from two autistic subjects (C, D). Yellow lines delineate scan path. Diameter of red circles is scaled proportionally to the fixation time in that region of the face.



Typical Viewer



Atypical Viewer



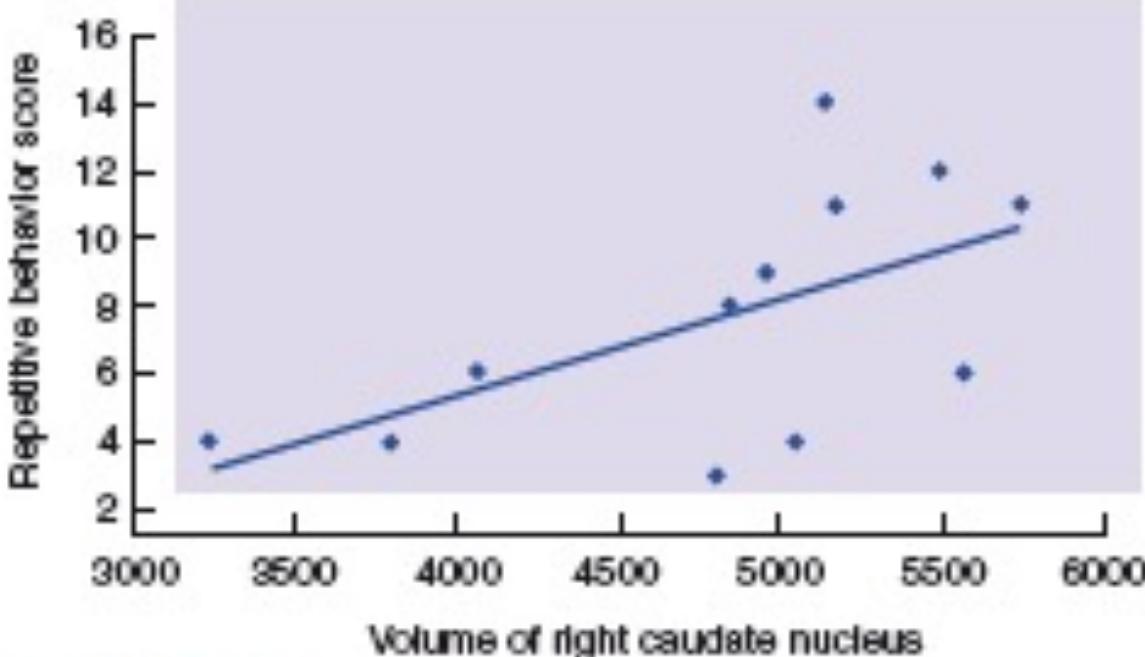
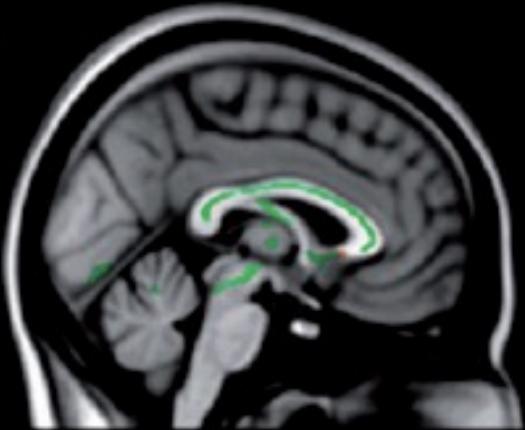
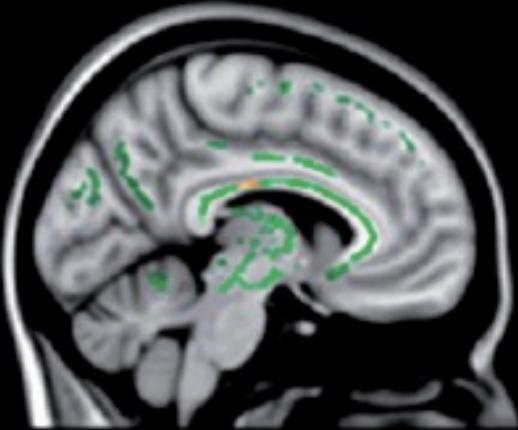
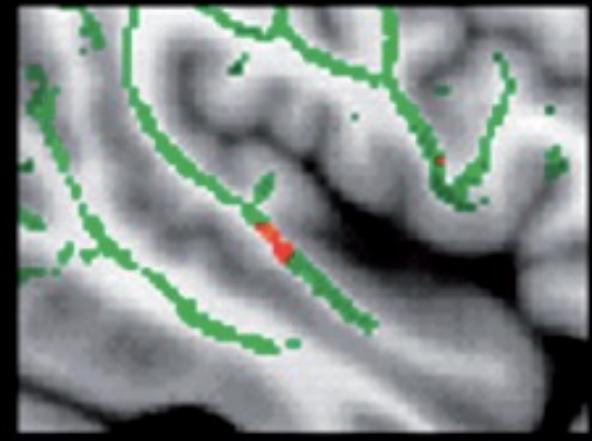
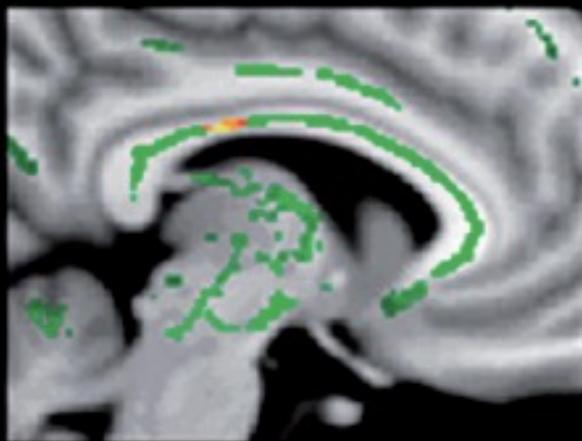
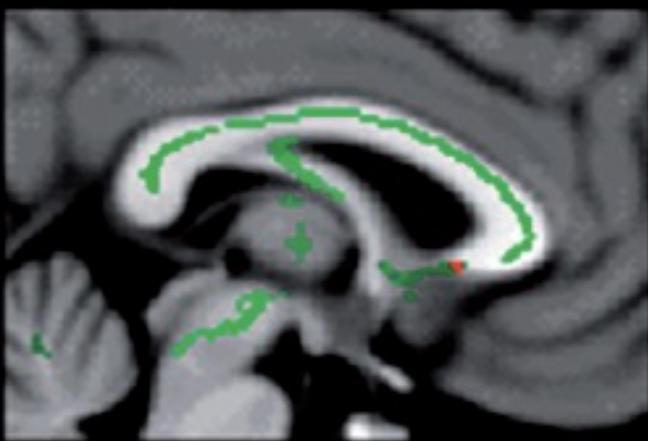
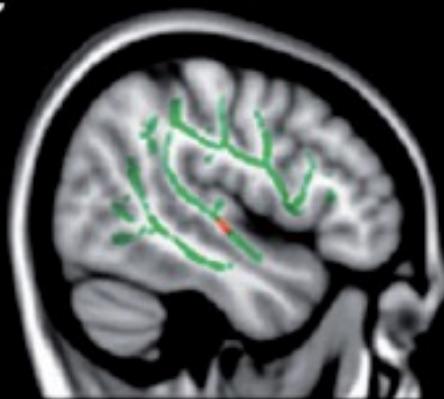


FIGURE 17.7 Caudate Nucleus and Stereotyped Behavior in Autism

The graph shows repetitive behavior scores of people with autistic spectrum disorders as a function of the volume of the right caudate nucleus. Larger volumes are associated with higher scores.

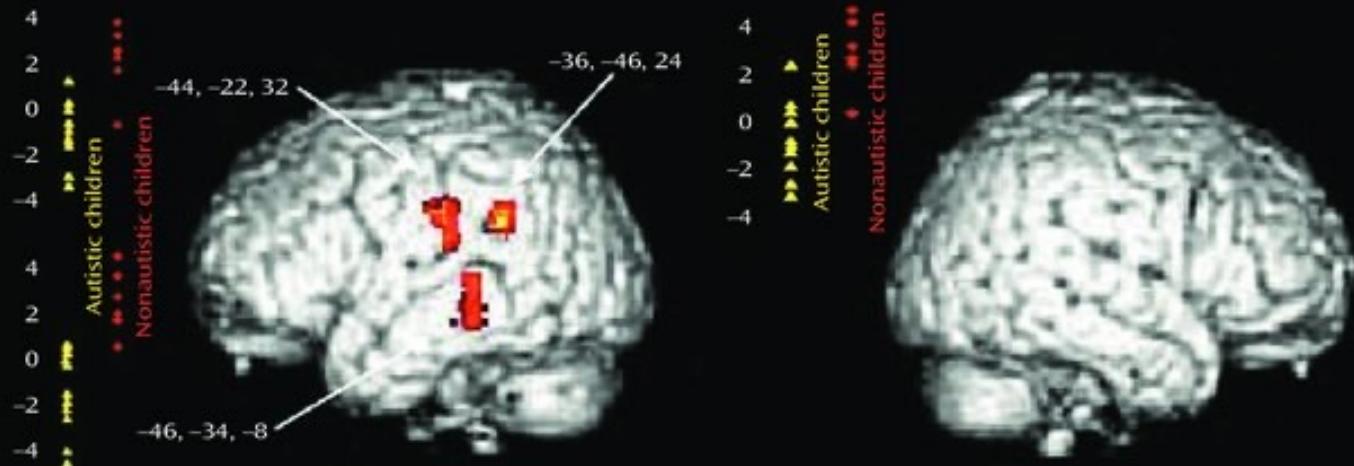
(Adapted from Hollander et al., 2005.)

Ruolo del nucleo caudato e stereotipie

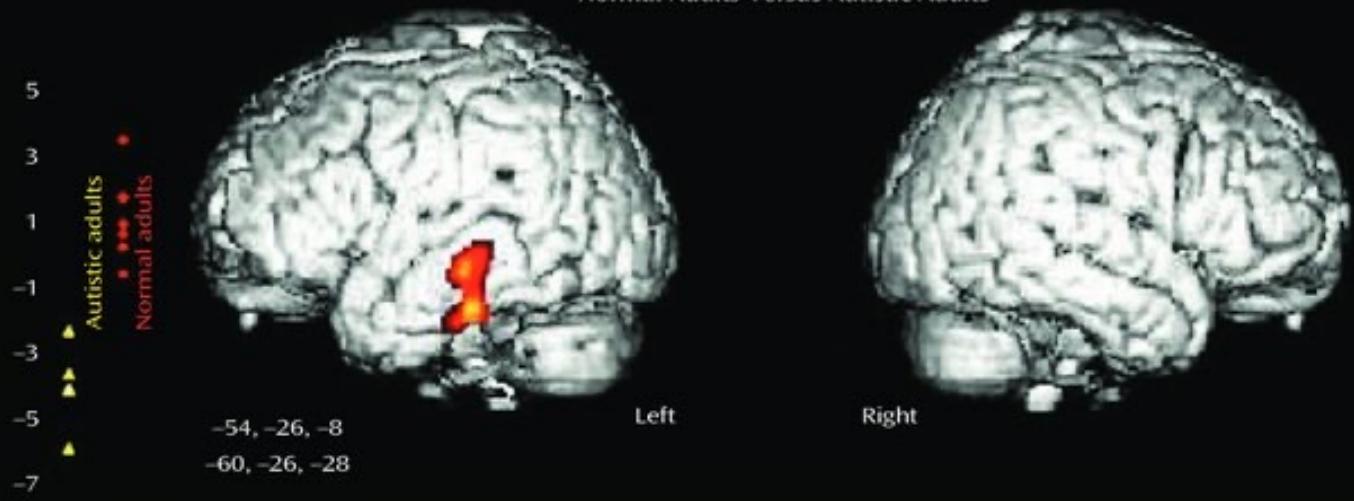
A**B****C**

Ridotta interconnettività negli autistici

Nonautistic Children Versus Autistic Children



Normal Adults Versus Autistic Adults



**Perception of Complex Sounds in Autism:
Abnormal Auditory Cortical Processing in Children**