

Lecture 4

Individual Differences in Cognitive Development

Developmental Psychopathology

- Links study of typical development to study of developmental disorders
 - Abnormality is normal
 - we all can act oddly
 - Disability is changeable
 - disability level can change over time
 - Adulthood may be better or worse
 - e.g. may overcome disability or struggle more as adult level skills become more important
 - Diagnosis depends on the social context
 - DSM
 - Psychopathology may reside “..not in the individual, but in the adaptiveness of the relationship between the individual and context.”

(Sameroff and McKenzie, 2003, p.613)

Chromosomal and Genetic Abnormalities

- Provide insight into complexities of nature and nurture
 - Knowing their origins helps limit the effects
 - Information can help the families
 - Information can help address related prejudice
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- Every cell with a nucleus in the human body has 46 chromosomes
 - Exception sperm and ovum (*gametes*)
 - only 23 chromosomes
 - combine to form an entirely new 46 chromosome cell called a *zygote*

Chromosomal and Genetic Abnormalities

- Suggested that one of every 200 births - live infant is born with 45, 47, or more rarely 48 or 49 chromosomes
- Results in specific syndromes (clusters of distinct characteristics that tend to occur together).
- Usually the cause is **three** chromosomes instead of two at a particular place - this is called a **trisomy**

Chromosomal and Genetic Abnormalities

Down Syndrome – most common cause of mental impairment

1 in 800/1000 births

- Trisomy-21: has three copies of chromosome 21
 - Not inherited – occurrence can appear random
 - 300 distinct characteristics can result from trisomy-21
 - Difference in symptoms and severity between individuals with Down syndrome
 - Several parts of the brain affected
 - Reduced brain size
 - Reduced number and size of neurons
 - Abnormal dendrites
- (Capone, 2001)*
- Brain pathology almost identical to that found in Alzheimer's Disease
 - higher risk of developing Alzheimer's Disease (around 35 – 40 years) further affecting communication skills

Chromosomal and Genetic Abnormalities

Down Syndrome - Cognition

- Impairment ranges from moderate to severe *(Moldavsky et al, 2001)*
- Cognitive spurts and regressions
- Rate of development slows throughout childhood and adolescence *(Capone, 2001)*
- Later adulthood – dementia impacts upon measures of Down Syndrome development
- Most acquire speech, but this is delayed *(Cohen, 2005)*
- Expressive language more impaired than comprehension
- Deficits in short-term memory and auditory processing *(Laws and Gunn, 2004)*
- Visual-spatial abilities relatively good

Chromosomal and Genetic Abnormalities

Fragile X syndrome - second most common cause of mental impairment

1 in 4000 births (male) - most common inherited cause

1 in 8000 births (female)

- Carried on X chromosome – males more likely to be affected
- Over-replication of a triplet of DNA nucleotides (*cytosene, guanine, guanine*) during conception

General population	between 6 and 50 replications
Carriers of Fragile X	between 50 and 200 replications
Fragile X manifested	over 200 replications

- Over 200 replications: FMR-1 gene no longer active
 - Adversely affects synaptic maturation, brain circuitry (*Reiss and Dant, 2003*)
 - Structural abnormalities found in several brain areas

Chromosomal and Genetic Abnormalities

Fragile X syndrome – Cognition

- Language skills: plateau at around 4 years
 - Cognitive and adaptive behaviour begins around 5 years and plateaus by late childhood/early adolescence
(Reiss and Dant, 2003)
 - Notable weaknesses in:
 - Visuo-spatial cognition
 - Sequential information processing
 - Motor coordination
 - Arithmetic
 - Executive functions
 - Relative strengths:
 - Verbal long-term memory
 - Acquired information
- (Volkmar and Dykens, 2002)
- Fragile X males: Most have mental deficiency – moderate to severe
 - Fragile X females: Many fewer and tends to be mild (second X chromosome protects (King et al, 2005).

Chromosomal and Genetic Abnormalities

Williams Syndrome

1 in 20,000 births - rare

- Random mutation – small deletions of several genes on chromosome 7
(Karmiloff-Smith and Thomas, 2003)
- Typically associated with mild to moderate mental deficiency
- IQ scores typically in 50 – 70 range
- Problems also include cardiac and kidney problems, hypersensitivity to sound
- Disorder linked to visual system and frontal lobe
(Bower, 2004)

Chromosomal and Genetic Abnormalities

Williams Syndrome – Cognition

- Striking difference between visuo-spatial abilities and linguistic functioning
- Deficient short-term visuo-spatial memory
- Severely impaired visuo-spatial skills
 - e.g. in adolescence, unable to perceive gross differences in spatial orientation and impaired ability to copy stick figures
- Interested in human faces
 - Can infer 'wanting' and 'thinking' from facial cues

(Elgar and Campbell, 2001)

Chromosomal and Genetic Abnormalities

Prader-Willi Syndrome

1 in 15,000 births

- Genomic imprinting – genes are expressed (activated) depending on whether they have been inherited from mother or father
- Involves genes in a certain area of chromosome 15 that normally solely expressed by chromosome inherited from father (*Milner et al, 2005*)
- 70% of cases – deletion of these genes
- Most other cases both chromosome 15s inherited from mother
- Leading cause of death – hyperphagia (excessive eating) due to disturbance of hypothalamus

Chromosomal and Genetic Abnormalities

Prader-Willi Syndrome – Cognition

- Exactness, order, cleanliness and sameness
- Intellectual functioning ranges from borderline to moderate levels
- Average IQ 70 *(Hodapp et al, 2006)*
- Different genetic subtypes – research suggests possible different features

Paternal gene deletion:

- lower intelligence (especially verbal)
- More frequent or severe behavioural difficulties
- High ability to solve jigsaw puzzles

Maternal genetic condition

- Decreased performance on visuo-spatial tasks
- Fewer facial characteristics
- Tendency for severe depressive symptoms
- Impairments in social interaction

(King et al, 2005; Milner et al, 2005)

Chromosomal and Genetic Abnormalities

Phenyketonuria (PKU)

- More prevalent in Northern European countries (*Welsh and Pennington, 2000*)
- Double recessive genes for PKU in newborns results in severe mental deficiency if consume the phenylalanine – amino acid present in many foods
 - Amino acid build up in body fluids causing brain damage and progressive mental deficiency
 - Immediate diet change can result in normal or close to normal development
(Hillman, 2005)

Pervasive Developmental Disorders

Autistic Disorder – Diagnostic concept

- 1960s – a ‘rare and seriously handicapping disorder, usually associated with intellectual disability’ (*Rutter, 2005*)
- Later research findings – autistic-like abnormalities could arise in individuals of normal intelligence
- Milder, but similar conditions, though different, are now termed the ‘broader phenotype’
 - Such abnormalities found in around one fifth of first-degree relatives of people with autism

Phenotype: a group of observable characteristics of an individual that results from the interaction of its genotype with the environment

Pervasive Developmental Disorders

Autistic Disorder

- Kanner (1973): term 'Autistic'
 - Communication deficits
 - Good, but atypical cognitive potential
 - Behavioural problems e.g. obsessiveness, repetitive actions and unimaginitive play
 - Fundamental disturbance: an inability to relate to people and situations from the beginning life
 - 'Happiest when left alone', 'like in a shell', acting as if people weren't there'

Pervasive Developmental Disorders

- **Autistic disorder:** spectrum of severely disabling disorders related to autism

(Akshoomoff, 2006)

- Inborn defect impairs perceptual and cognitive functioning
- Precise causes unknown - difficult to understand and treat
 - Structural differences in the brain /neurological system *(Siegel, 1996)*
 - Fundamental disturbance of the central nervous system implicated

- Genetic liability:
 - Concordance 60% monozygotic twins/5% dizygotic twins
 - Higher 'broader phenotype' concordance in monozygotic than dizygotic
 - Support from family studies *(Rutter, 2005)*
- Between 3 and 12 genes suggested to be implicated for autism that act synergistically
- Explains why most siblings do not have ASD – do not have all of these genes

- Parental social class and country of origin
- Disproportionately likely to have parents of a high socio-economic background (*Kanner 1943*)

But, date of study/definitions of 'class'?

- Later studies – No real association.
Perhaps referral bias - higher rates of diagnosis
(*e.g. Fombonne et al, 2001*)

Pervasive Developmental Disorders

- **DSM classification and diagnosis** (APA, 2000):

A total of six or more from 1, 2 and 3...

1. Qualitative impairment in social interaction manifested by at least two of the following:
 - impaired non-verbal behaviour
 - failure to develop age-appropriate peer relationships
 - lack of spontaneous sharing of enjoyment or interests
 - lack of social or emotional reciprocity
2. Qualitative impairment in communication manifested by at least one of the following:
 - delay or lack of spoken language
 - when speech is inadequate, impairment in initiating or sustaining conversation
 - stereotyped, repetitive or idiosyncratic language
 - lack of age-appropriate, spontaneous make-believe or social imitative play
3. Restricted, repetitive, stereotyped patterns of behaviour, interests, or activities manifested by at least one of the following:
 - abnormal preoccupation with stereotyped and restricted interests
 - Inflexible adherence to nonfunctional routines or rituals
 - stereotyped and repetitive motor mannerisms
 - Persistent preoccupation with parts of objects

Pervasive Developmental Disorders

Autism: Research focus often on pre-school and school-age children

- Most parents of children with autism become concerned about symptoms by age 2
- Diagnosis generally occurs at approximately 5 years

Patterns of specific symptoms and their severity can be very different across people

Triad of Impairments

- Social interaction
- Communication
- Restricted, repetitive stereotyped behaviour and interests

Associated impairments

- Sensory/perceptual impairments
- Intellectual performance

Pervasive Developmental Disorders

Autism: Triad of Impairments

Social interaction

- Subtle differences in infancy/early years
 - Less likely to be visually responsive
 - Less likely to respond to their names
 - More likely to show aversion to being touched by another
 - Fail to track people visually
 - Avoid eye contact
 - Exhibit an 'empty gaze'
 - Fail to respond with emotional expression and positive affect
 - Show little interest in being held

(e.g. Werner et al 2000; Stone, 1997)

Pervasive Developmental Disorders

Social interaction

- Joint attention interactions: lower level of pointing and eye contact (shared experience)
- Impaired facial recognition/memorization ability (*Dawson and Toth, 2006*)
- Impaired recognition of facial expression
- Atypical facial processing – focus on mouth not eyes
- Lack of understanding of social cues
- Aloofness, disinterest and lack of reciprocity – lacks engagement with peers
-
- May have problems forming friendships throughout life

Pervasive Developmental Disorders

Communication

- Disturbed verbal and non-verbal communication – sometimes absence of speech
- Approximately 30% of children with autism never develop spoken language
(Dawson and Toth, 2006)
 - Language ability usually delayed or abnormal (tone, pitch, rhythm)
 - Echolalia (echoing back another speech)
 - Pronoun reversal (others referred to as I or Me, the self as He/She/Them/You)
(also noted in schizophrenia and blindness)
 - Irrelevant details, inappropriate shifts in topic, disregard of normal rules of conversation (or no conversation at all)
 - Hyperlexia – very good single word reading, but little comprehension
- But some do function highly – can read, tell stories and communicate better when prompted

Pervasive Developmental Disorders

Restricted, repetitive stereotyped behaviour and interests

- Inflexible adherence to ritual/routines *(Richler et al, 2007)*
- Frequency and severity of self-stimulation behaviours e.g. head banging
- rocking; walking on their toes, whirling; arm, hand or finger flapping *(Klinger et al, 2002)*
- Higher order repetitive , obsessive activities and interests – more common in older children
 - Unusual preoccupations with aspects of the environment (e.g. numbers/objects)
 - May compulsively collect items
 - Overly-preoccupied with hobbies
 - Motor routines (e.g. rearranging objects; mealtime/bedtime rituals)
 - Distressed by changes in the environment

Pervasive Developmental Disorders

Associated impairments

- Sensory/perceptual impairments
 - Oversensitivity to stimulation *(Dawson and Toth, 2006)*
 - Sensory input – disliked, feared or avoided
 - Under-sensitivity to stimulation
 - Failure to respond to sounds, verbal communications or sight of others *(Rogers and Ozonoff, 2005)*
 - Contradiction
 - May ignore loud clock noise , but be fascinated by a quite watch ticking *(Volkmar and Klin, 2000)*
 - Overselectivity
 - Obsessive attention to a particular stimulus, while ignoring competing stimuli
 - Failure to attend to range of stimuli affects child's learning

Pervasive Developmental Disorders

Intellectual Performance

- Wide range of intelligence in autism, including above average
- 70-75% - mental deficiency. Many youth severe or profound deficiency
- IQ – important difference between people with autism
 - Higher IQ: less severe autistic symptoms and a greater chance of normal functioning in later life (Howlin et al, 2004)

Pervasive Developmental Disorders

Intellectual Performance

- Generally - uneven cognitive development.
- Deficits in...
 - Abstract and conceptual thinking
 - Language development
 - Social understanding
- Relative strengths...
 - Rote learning
 - Rote memory
 - Visuo-spatial skills

Pervasive Developmental Disorders

Intellectual Performance

- Performance scores usually better than verbal scores across the IQ range
 - Can lessen from preschool to adolescence if higher functioning
 - perhaps because language skills improve *(e.g. Joseph et al, 2002)*
- Autistic symptoms with such wide variability in general intelligence puzzling

BREAK



15 MINUTES!

Also – Savant abilities - small minority of youth exhibit ‘splinter skills’

- Much higher level than expected on basis of their general intelligence
- Much higher level than normally-developing youth

(Miller, 1999)



Nadia, age 3



Stephen Wiltshire

Pervasive Developmental Disorders

- Adaptive Behaviour
 - Difficulties dealing with aspects of everyday life
 - Vineland Adaptive Behaviour Scales
 - Distinct profile for people with autism when comparing with typical peers (Kraijer, 2000)

Self-help and daily living skills	As expected on basis of mental ability
Communication skills	Fall below mental ability
Social skills	Far below expected level

- Other features include tantrums, aggression, hyperactivity and self-injurious behaviour – not specific to autism, but can interfere further with functioning

Pervasive Developmental Disorder

Underlying Cognitive Deficits

Autism and Theory of Mind

- Do children with autism have an understanding of minds?
- We usually realise that
 - People have different mental representations (beliefs) about the world than we do
 - People can have a representation about the world that is inaccurate
(a **false belief**)

Difficulty comparing ASD children and typically developing children due to the need for different chronological ages - e.g. Typical – 6yrs / ASD – 12 years – because of the difference in life experience

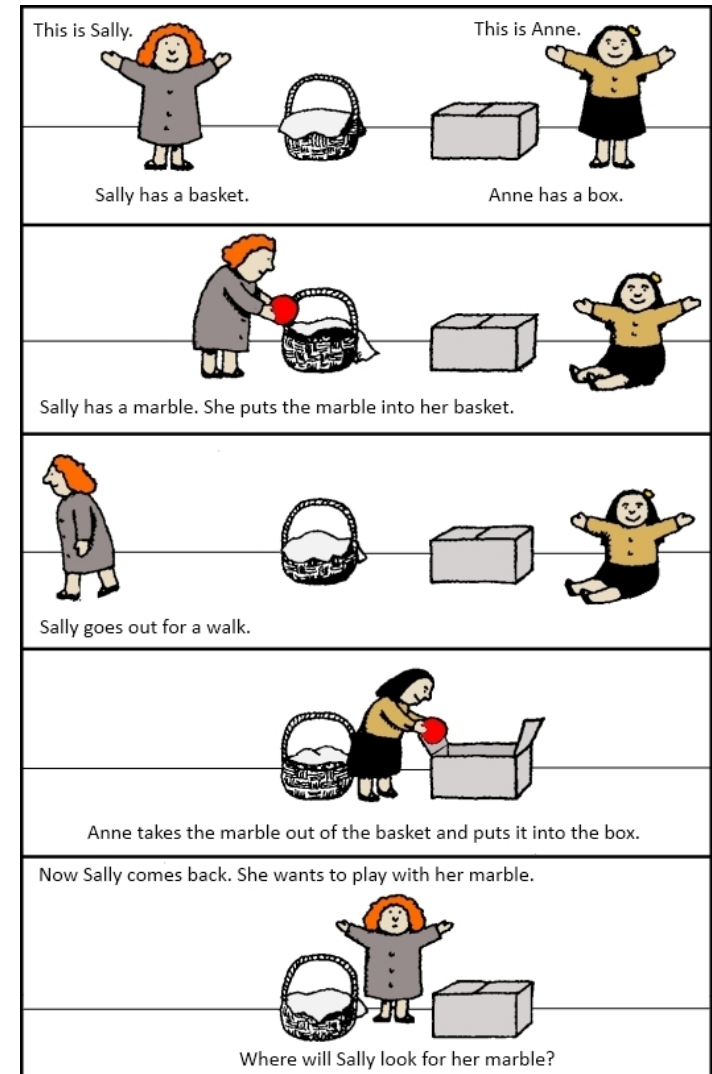
- Experiments tend to include comparisons with children with other developmental disorders

Pervasive Developmental Disorders

- Baron-Cohen et al (1985)

Sally-Anne false belief test

- Typically developing children aged 4 years
- Children with Autistic Spectrum Disorder with mental age over 4 years
(age of success on f-b test for typically developing children)
- Down Syndrome children with mental age of 4 years



Pervasive Developmental Disorders

Findings

- 80% 'typical' children and 80% Down syndrome children succeeded at task
- Only 20% Autistic Spectrum children succeeded
- Unlikely to be due to general learning difficulties, as children with Down Syndrome were successful
- ASD children - problems understanding that another person may have a false belief

Pervasive Developmental Disorders

- Baron-Cohen (1986)
- Asked children to reassemble picture stories (4 pictures in each)
 - **Mechanical** story – no people
e.g. a balloon blowing away and bursting on a tree
 - **Behavioural** story – included people but did not require an understanding of what they were thinking
e.g. boy going into shop and buying sweets
 - **Mentalistic** story – required an understanding of the beliefs about the characters in the pictures
e.g. mother eats last biscuit from box while boy out playing
- ASD children
 - Success at mechanistic and behavioural stories

Pervasive Developmental Disorders

- Findings
 - ASD children
 - Success at mechanistic and behavioural stories
 - Mentalistic stories
 - pictures put in jumbled order.
 - could not explain why boy looked surprised when he came in and opened the box

Asperger's Syndrome

- Hans Asperger (1944)
- Many behaviours similar to autism
- Usually appears later than in other ASDs
- Substantial long-term disability
 - Social impairment, restricted interests, repetitive behaviours
 - 'Eccentric', 'odd' behaviours – nonverbal learning disability, need for sameness, physical awkwardness, social skills issues, emotional instability
 - Not associated with severe delay in language development, social interactions or age-appropriate self-help skills

- **Asperger's Syndrome**

- Linked to:

- Severe, sustained impairment of interpersonal interactions (facial expression, body postures, gestures)
- Failure to develop peer relationships
- Higher rate of suicide during adolescence than among typically developing adolescents
(e.g. *Fitzgerald, 2007*)

Pervasive Developmental Disorders

Aetiology of Autistic Spectrum Disorders

- Genetics
 - Tiny and rare variations in genes increase risk of autism Spectrum Disorder (ASD) (*Autism Genome Project, 2007*)
 - 'Faulty wiring' – glutamate neurotransmitter system
- Often co-occurs with PKU and fragile X syndrome (*Smalley, 1998*)
 - Implies a genetic link
- Twin studies – concordance rates
 - MZ twins: between 60 and 91% DZ twins: between 0 and 9%
- Triad of impairments: social impairments, communication impairments and repetitive behaviours show high levels of heritability (*Ronald et al, 2006*)

Pervasive Developmental Disorders

- Chromosomes
- Link with a number of chromosomal disorders including tuberous sclerosis and fragile X syndrome
- Autistic disorder linked to duplications, deletions and inversion of chromosomes
(e.g. Wolpert et al, 2001)
- But accounts for relatively few instances (fewer than 10%)
(Folstein and Rosen-Sheidley, 2001)

- Biochemical factors
- Neurotransmitters
- Early studies suggested low levels of dopamine and serotonin – essential for effective cognitive, behavioural and motor functioning and mood regulation (e.g. Ernst et al, 1997)
- But research has been inconclusive – methodological issues
 - e.g. small samples sizes, different research methodologies, experiment control issues

- Perinatal factors

- Prenatal factors (e.g. rubella during pregnancy, intrauterine exposure to certain drugs, depressed maternal immune functioning) generally found in individual case reports – probably account for small percentage of cases of ASD *(Frombonne, 2002)*
 - e.g. rubella almost eradicated from Western countries

- Postnatal events

- Recent studies failed to identify link between infectious diseases in first two years of life and autism
 - e.g. inflammatory bowel disease, MMR vaccine *(Wakefield et al, 1998)*

- Brain Abnormalities
- Autopsy studies, fMRI and EEG studies – evidence of link between abnormal brain development and autism
- Autopsy studies
 - Limbic system – smaller and more dense than normal, dendrites shorter and less well-developed
 - Cerebellum – associated with motor skill deficits (e.g. balance, dexterity and grip)
 - Overly large brain size and enlarged ventricles in the brain

- fMRI

- Frontal lobes, limbic system, cerebellum, basal ganglia (*Sokol and Edwards-Brown, 2004*)
- Confirm larger brain size and poorer neural connectivity
- Lack of theory of mind – linked to decreased activation of prefrontal cortex and amygdala (*Castelli et al, 2002*)

- EEG

- Abnormal EEG pattern in frontal and temporal lobes, some clinical seizures (*Dawson et al, 1995*)

- Treatment and Care - Difficult

- Resistance to change, lack of response to outside world, lack of interest in reinforcers, over-selective attention (learning situation specific?)

- Drug treatments:

- Antipsychotic medications most common to help control behaviours
 - Can reduce repetitive behaviours, social withdrawal, aggression and challenging behaviour (hyperactivity, tantrums, mood swings, self-abuse)

But:

- Not all children responsive to these drugs.
 - Side effects include dizziness, sedation, appetite increase, dyskinesias

(Campbell et al, 1997)

- Behavioural training methods:
- Basic self-help, social and communication skills
 - **Modelling** and **Conditioning** e.g. reinforcement of eye-contact, toileting behaviours, play with peers...
 - Sign language if required
- Few assessments of the efficacy of training methods, but may be the most effective way of promoting social functioning over the long-term
- Time-consuming and repetitive
 - Need to train parents
 - Child learns appropriate behaviours for familiar environments

- Inclusion strategies:

- Help with important life skills and transitions e.g. finding and maintaining employment – ‘supported employment’ (supports employee and employer)
- Increases rates of employment *(Mawhood and Howlin, 1999)*
- Increases integration, satisfaction and self esteem *(Kilsby and Beyer, 1996)*

- Asperger's specific: Treatment

- Psychotropic medication (antidepressant, antipsychotic)
- Mood stabilizers (e.g. lithium)
- Behavioural treatments e.g. social-behavioural learning strategy intervention (Bock, 2007)

Reduces problem behaviour, improves co-operative learning activities with peers



Chris Packham



Courtney Love



Daryl Hannah



Vincent D'onofrio



Dan Aykroyd

Reading

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