Types of developmental disorder

1. Disorders caused by well-understood genetic abnormalities (e.g., Down syndrome)
2. Disorders defined by behavioural deficit (e.g., dyslexia, SLI, autism)
3. Developmental disability of unknown aetiology
4. Disorders caused by environmental factors (e.g., impoverished environment, Foetal Alcohol Syndrome)

Genetic syndromes

- Alagille syndrome
- Angelman syndrome
- Apert syndrome
- Beddworth-Wiedemann syndrome
- Bloom syndrome
- Branchio-oto-renal syndrome
- Cri du chat syndrome
- Crouzon syndrome
- Di-George syndrome
- Down syndrome
- Ehlers-Danlos syndrome
- Fragile-X syndrome
- Jackson-Wales syndrome
- Jervell-Lange-Nielsen syndrome
- Kallman syndrome
- Kearns-Sayre syndrome
- Langer-Giedion syndrome
- Leesch-Pylén syndrome
- Li-Fraumeni syndrome
- Marfan syndrome
- Miller-Dieker syndrome
- Nijmegen breakage syndrome
- Pfeiffer syndrome
- Prader-Willi syndrome
- Romano-Ward syndrome
- Rubinstein-Taybi syndrome
- Smith-Magenis syndrome
- Stickler syndrome
- Treacher Collins syndrome
- Treto-Piloni-phenotypic syndrome
- Turner syndrome
- Usher syndrome
- Waardenburg syndrome
- WAGR syndrome
- Williams syndrome

Explanatory frameworks

- Static classical cognitive neuropsychology (e.g., Temple, 1997)
- Neuroconstructivist (e.g., Karmiloff-Smith, 1998)
Classical (static) cognitive neuropsychology

- For disorders with uneven cognitive profiles (e.g., in adulthood), use selective deficits to inform normal structure (general deficits less interesting...)
- Temple (1997):
  - "The objective of case studies in cognitive neuropsychology is not to produce a clinical taxonomy, but to propose selective deficits of a common modular architecture of a developmental system"
  - "It may be common for children to have problems with several components of the system, but the most informative child, in terms of revealing the structure of the system, will be the child who has selective difficulty with a particular element and therefore a dissociation of skills"

Temple (1997):
- "Downstream effects do not mean that a developing system cannot have a modular organisation, but they may make the appearance of the classical double dissociations, which are the hallmark of cognitive neuropsychology, difficult to attain within developmental disorders"
- "If the brain reorganised and generated new modules then abnormal performance would not necessarily reflect a normal system minus those components that are disrupted, which is a basic assumption of cognitive neuropsychology (Saffran, 1982)"
- "From the perspective of cognitive neuropsychology, which has little interest in the issue of biological localisation, it is of no consequence whether language develops in the right or the left hemisphere, provided that the functional architecture of the language system is the same in both cases"

Problems with static approach

- Use of static models eliminates the developmental process, a core causal element
- Functional modules are an outcome of the developmental process, not a precursor in domains such as language, face processing, executive function, imaging studies show increasing selectivity, localisation, and lateralisation of activity with age
- Behavioural impairments = outcome of a process of development involving a long history of interaction with the environment
- Coarse standardised test scores showing performance in the normal range cannot reveal if underlying processes are atypical
- Gene expression in the developing brain is typically widespread
- Developmental process involves compensation and interaction - unlikely that deficit in one module will not affect others during development ("Residual Normality")
- Selective deficits often explained using non-developmental models

Gene expression in the development of neocortex: plaid or patchwork?

The relation of genes to cognition

- Genetic effects usually more widespread than specific part of brain eventually specialised for single cognitive function
- Genetic effects tend to modulate
  1. Brain size (no. neurons or synapses)
  2. Neuronal migration
  3. Neurotransmission
- Grammar-specific gene doesn’t look plausible...

Problems with the static approach

- Examples of apparently selective developmental deficits explained using non-developmental models
Arithmetic
Developmental Dyscalculia
(case study SW)

Face Processing
Developmental prosopagnosia
(case study AB)

Face Processing
Developmental prosopagnosia
(case study Dr S)

Spelling
Developmental dysgraphia
(case study AH)

Static models
- Static models (usually adult) - not bad place to start to generate hypotheses about source of developmental deficits
  - But final explanation must be in terms of way specialised components develop
    - e.g., modules of reading system - how do they know what to do? Reading is not even innate

Neuroconstructivist approach: Karmiloff-Smith (1998)
- Disorders are the result of development guided by atypical neurocomputational constraints from embryogenesis onwards
- Causes will be in terms of low-level neural properties + developmental process
  - e.g. firing properties of neurons or local connectivity
Neuroconstructivist approach: Karmiloff-Smith (1998)

- Approach predicts both strengths and weaknesses in behavioural outcome
  - Small differences in initial start state can lead to divergent behavioural outcomes
  - Apparently normal behaviour in a disorder may have different cognitive processes underlying it

- Rejects innate modularity - “different domain-relevant mechanisms become domain-specific through ontogeny”
- Empirical approach traces back developmental trajectories to infancy
- Uses sensitive measures to explore nature of underlying processes
- Takes developmental computational neuroscience perspective
  - Includes brain imaging data
  - Employ developmental computational modelling

Problems with neuroconstructivism

- Risks rejecting possibility that anything can develop normally in a genetic developmental disorders
- Not much empirical evidence that developmental disorders show radically different modular structure (though methodological issues here)
- Difficulty of assessing functional significance of atypicalities in brain structure / metabolism / processing

Developmental disorders of language

- Importance of plasticity illustrated by fact that children who experience same brain damage as adult aphasics end up with no symptoms of aphasia (Bates & Roe, 2001)
- Whatever is wrong in the developmentally disordered brain, plasticity cannot overcome it
- Doesn’t imply plasticity isn’t trying... plasticity itself may be atypical

Developmental disorders of language

- Auditory agnosia
  - Landau-Kleffner syndrome (Acquired Epileptic Aphasia)
    - Profound receptive language impairment (may extend to total disappearance of auditory verbal comprehension)
    - Onset 18 months - 13 years (peak incidence 4 years)
    - 60% cases also seizure disorder with bilateral EEG abnormality
    - Disruption of processing of auditory input to Wernicke’s area?
    - No evidence of deafness - failure of words to be associated with meanings
    - Knock-on effects in development of syntax
    - Later the onset, better final language development

- Semantic disorders
  - Children with word finding difficulties
  - Semantic but not phonological errors in naming
  - Slow naming times
  - Impoverished word definitions

- Grammatical disorders
  - SLI - esp. errors of morphology (differ across languages)
  - KE family (motor deficits too - FOXP2 gene)
  - Grammatical SLI - restricted to difficulties with representational dependencies within syntactic constructions
Three current theories of SLI

1. Deficits to rule-based language-specific structures (e.g., van der Lely)
   - Impairment in specific structural relationships (agreement, specifier head-relations)
   - Absent linguistic features
   - Fixation in a period of development where tense marking is "optional"
   - Problems in more general language functions (implicit rule learning, representing relationships between structures)

2. Non-linguistic processing deficit that particularly impact on language (e.g., Tallal)
   - Reduced processing rate
   - Capacity limitations on cognitive processing
   - Deficit that particularly affects phonology
   - Low-level perceptual or temporal processing deficit

3. Procedural-Declarative theory (e.g., Michael Ullman)
   - Grammar relies on procedural memory, vocabulary on declarative memory
   - SLI = developmental deficit to procedural system

Developmental disorders of language

- Developmental dyslexia
  - Problems in forming phonological representations exist prior to literacy (e.g., revealed by phoneme discrimination, onsets-rhyme knowledge)

- Pragmatic disorders
  - Language difficulties overlap with wider social communicative difficulties (autistic spectrum)

Other disorders

- Williams syndrome - language relative strength compared to visuospatial skills
- Downs syndrome - language development much delayed (esp. syntax), poor phonological working memory
- Autism - varying levels of language development, particular pragmatic impairment

Other disorders

- Non-Verbal Learning Disability (Rourke, 1987, 1989)
  - Widespread brain damage causes recurring pattern:
    - Relative weaknesses
      - Bilateral tactile-perceptual deficits, more marked on the left side of the body
      - Impaired visual recognition and discrimination
      - Impaired visuospatial organisation
      - Bilateral psychomotor co-ordination problems, more marked on left
      - Difficulties managing novel information
    - Relative strengths
      - Simple motor skills
      - Auditory perception
      - Rate learning
      - Selective and sustained attention for auditory-verbal information
      - Basic expressive and receptive language
      - Word reading and spelling

The example of Williams syndrome

- Rare genetic disorder (1 in 20,000 live births) caused by a deletion of 16 genes on the long arm of chromosome 7 at q11.23
- Clinical features:
  - Heart abnormalities - typically SVAS
  - Facial dysmorphism
  - Small stature
  - Hemias
  - Hoarse voice
  - Premature ageing of skin
  - Constipation
  - Hyperacusis
  - Abnormal gait
Cognitive features:
- low IQ
- a specific personality profile ('hypersociability', empathy, anxiety)
- poor visuospatial constructive skills
- particular difficulty with number processing
- relatively good language abilities (though developmental delay)
- relatively good face processing abilities

Brain anatomy in WS
- 80% of normal brain volume
- anterior regions and cerebrum: small in proportion to other brain regions
- limbic and frontal regions: small, but proportionally normal to other brain regions
- cerebellum: large in proportion to other brain regions
- malformations in dorsal regions
- total grey matter reduced
- abnormal layering, orientation, density and size of neurons in several brain regions

Brain chemistry in WS
- Normal ratios in cerebellum:
  - Cho/Cre
- Abnormal ratios in cerebellum:
  - Cho/NAA
  - Cre/NAA
- Correlations with cognitive ability:
  - Increase in cognitive performance associated with increase in NAA for all cognitive tasks (particularly general speed of processing)

Structure-function relationships?
- Quote illustrates neuroconstructivist perspective on importance of substrate
- Possible response is to argue only some of atypicalities have functional importance (ungrounded)

Williams syndrome

- LOSS OF ~20 GENES?
Rossen et al. (1994): WS "presents a remarkable juxtaposition of impaired and intact mental capacities...[...]. Linguistic functioning is preserved in Williams syndrome while problem solving ability and visuospatial cognition are impaired."

"Although their IQ is measured at around 50, older children and adolescents with WS are described as hyperlinguistic with selective sparing of syntax, and grammatical abilities are close to normal in controlled testing. This is one of several kinds of dissociation in which language is preserved despite severe cognitive impairments, suggesting that the language system is autonomous of many other kinds of cognitive processing." (Pinker, 1994)


**Early views**

- Rossen et al. (1994): WS “presents a remarkable juxtaposition of impaired and intact mental capacities...[...]. Linguistic functioning is preserved in Williams syndrome while problem solving ability and visuospatial cognition are impaired.”
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**WS language: More recent research**

- Overall profile
  - Delayed language development (~ 2 years)
  - Usually MA / DS comparison groups - No independence from general cognition
- Precursors
  - Delayed pointing, impaired triadic interactions
  - Delay in using labels to aid categorisation
- Speech processing
  - Anomalous auditory ERPs
  - Lexical segmentation delayed
  - Phonological STM relative strength

**More recent research suggests:**

- Vocabulary acquisition
  - Vocabulary spurt doesn't coincide with usual semantic markers
  - Lexical constraints different? (whole object, taxonomic)
  - Difficulty in spatial, perhaps ‘relational’ vocabulary
- Semantics
  - Poorer / slower but not atypical in its underlying dynamics
  - Integration with syntax may be anomalous
  - Knowledge remains more perceptually-based and insufficiently abstract

**More recent research suggests:**

- Relation of grammar to vocabulary
  - Initially MLU predicts grammatical complexity
  - Complex structures apparently mastered
  - Grammar generally behind vocabulary (TROG vs. BPVS)
  - Exaggerated difficulty with complex structures
  - Atypical errors found in Italian, Spanish gender agreement, morphology, preposition use
- Pragmatics
  - Persistent deficits, e.g. in non-literal language (despite usage of this language)

**Overall picture in WS**

- Different lexical constraints
- Subtle grammatical impairments
- Atypical balance of semantics/phonology
- Language is relatively good but develops atypically

However, some researchers still prefer static view

- “the linguistic performance of [individuals with] WS can be explained in terms of selective deficits to an otherwise normal modular system” (Temple & Clahsen, 2003)
- (cf. inflectional morphology debate)
Comparison of disorders

- Comparison of developmental disorders may be informative about constraints acting on normal language development
  - e.g., importance of various information sources

Comparison of disorders

- Two types of information from patterns:
  - Function words - mark syntactic structure; fast automatic use associated with emergent left lateralisation of language (ERP marker)
  - Morphological analysis of words into component parts
    - [I scrambled the eggs] = [I scramble + ed the egg + s]

Comparison of disorders

- McDonald (1997): 3 routes to acquiring linguistic structure
  - Prosodic and/or phonological information used to segment input into meaningful units
    - [scrambled the eggs] = [I scrambled the eggs]
  - Analysis of distributional and co-occurrence patterns of linguistic elements

Acquisition of linguistic structure in different populations (McDonald, 1997)

<table>
<thead>
<tr>
<th>Routes to acquiring linguistic structure</th>
<th>Function words</th>
<th>Morphological decomposition</th>
<th>Comments</th>
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<tbody>
<tr>
<td>Successful Language Learners</td>
<td></td>
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<tr>
<td>Down's Syndrome</td>
<td>Unsuccessful</td>
<td></td>
<td></td>
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<tr>
<td>- Good vocabulary, good grammatical command</td>
<td>Language Learners</td>
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<tr>
<td>- Good morphology</td>
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<tr>
<td>- Good phonology</td>
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Unsuccessful Language Learners

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Diagnostic and phonological information</th>
<th>Function words</th>
<th>Morphological decomposition</th>
<th>Comments</th>
</tr>
</thead>
<tbody>
<tr>
<td>Down's Syndrome</td>
<td>Poorer auditory sensitivity, delayed onset of speech, difficulty in distinguishing sounds</td>
<td>Jargon-like speech</td>
<td>Decomposition and surface decoding problems</td>
<td>Poor auditory processing, delayed on-line processing / phonological information</td>
</tr>
<tr>
<td>Autism</td>
<td>Good vocabulary, good grammatical command</td>
<td>Language Learners</td>
<td></td>
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<tr>
<td>- Good morphology</td>
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Conclusions

- Developmental deficits in language not due to brain damage analogous to adult case
- Genetic developmental disorders can show auditory, semantic, grammatical, and pragmatic deficits
- Explanation in terms of atypical neurocomputational constraints operating during development + developmental process
- Detailed testing required to reveal atypical constraints
- Computational modeling required to reveal role of developmental process
- Use of non-developmental models still widespread - can play role
In a developmental disorder, cognitive mechanisms should never be described as \textit{INTACT/PRESERVED/NORMAL} vs \textit{IMPAIRED}.

The correct terminology is \textit{DEVELOPS NORMALLY} vs \textit{DEVELOPS ATYPICALLY (TO PRODUCE IMPAIRED PERFORMANCE)}.

Only \textit{behaviour} can be INTACT vs IMPAIRED.