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Dyslexia: a research update

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SASC Conference
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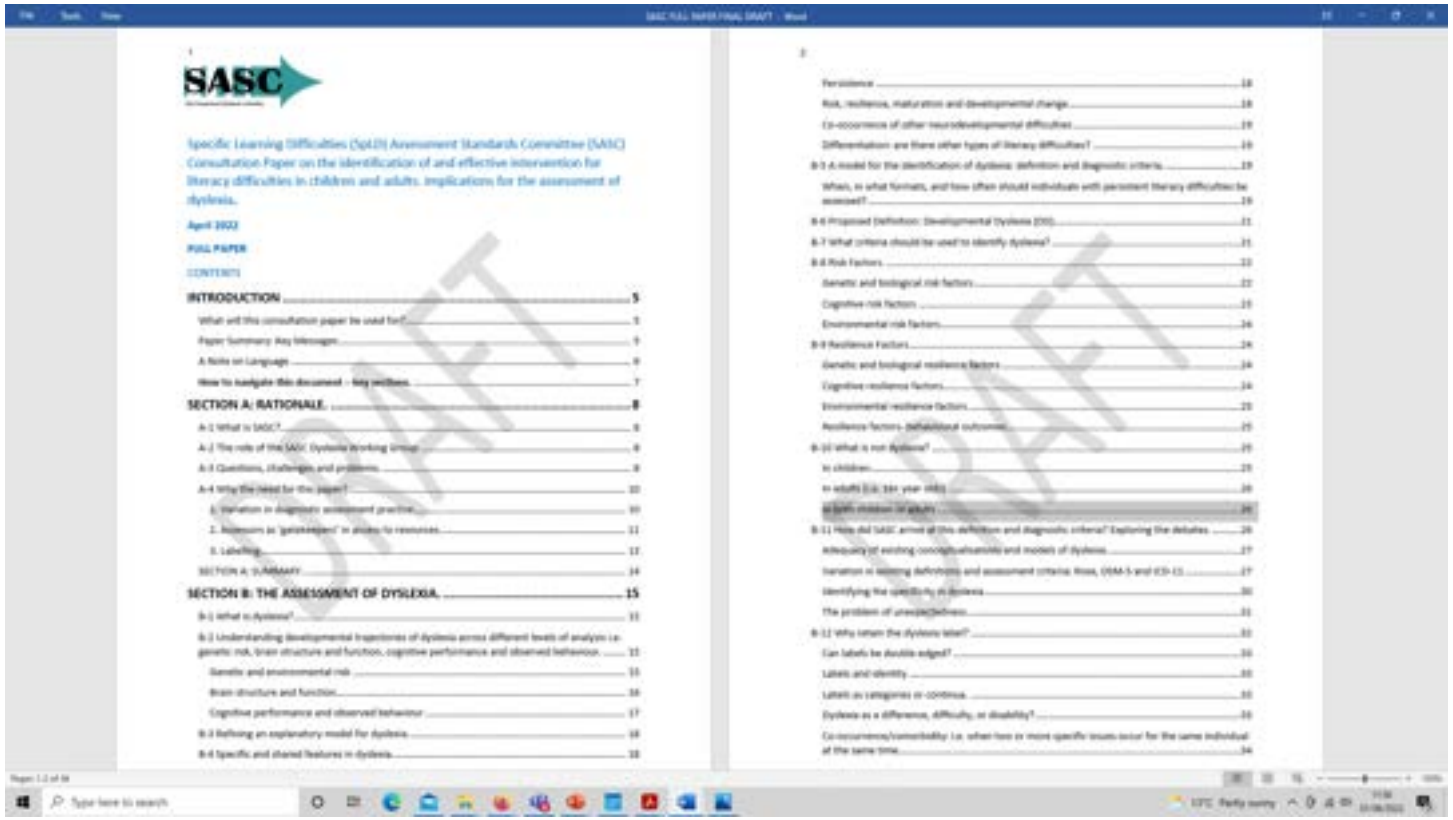
Remit of the talk

- Section B of SASC's recent consultation paper on dyslexia
  - conceptualisations of dyslexia
  - criteria for its identification.
- concepts such as persistence, dimensionality, risk, resilience, developmental trajectory and risk accumulation, underpinning a dynamic, dimensional model for understanding developmental difficulties, including dyslexia.



A (partial) history of Dyslexia definitions

- 'Congenital word-blindness' (Pringle-Morgan, 1896)
  - First medical description of reading-specific disability (case-study)
- World Federation of Neurology (Critchley, 1970)
  - Definition based on diagnostic criteria, primarily exclusionary
  - 'specific reading difficulty'
- International Dyslexia Association (1994)
  - Core cognitive deficits as inclusionary criteria (e.g., in phonological awareness and processing)
- DSM-IV (APA, 2004)
  - Reading Disorder defined by 'unexpected difficulties' in literacy, distinguished from those explained by other potential causes.
- Rose Review (2009)
  - UK working definition, focusing on functional educational deficits (not rigorous diagnostic criteria) and multiple mechanisms
- DSM-V (2013)
  - 'Dyslexia' embedded under 'Specific Learning disorder'; independent from 'Communication disorders' (for e.g., in language)

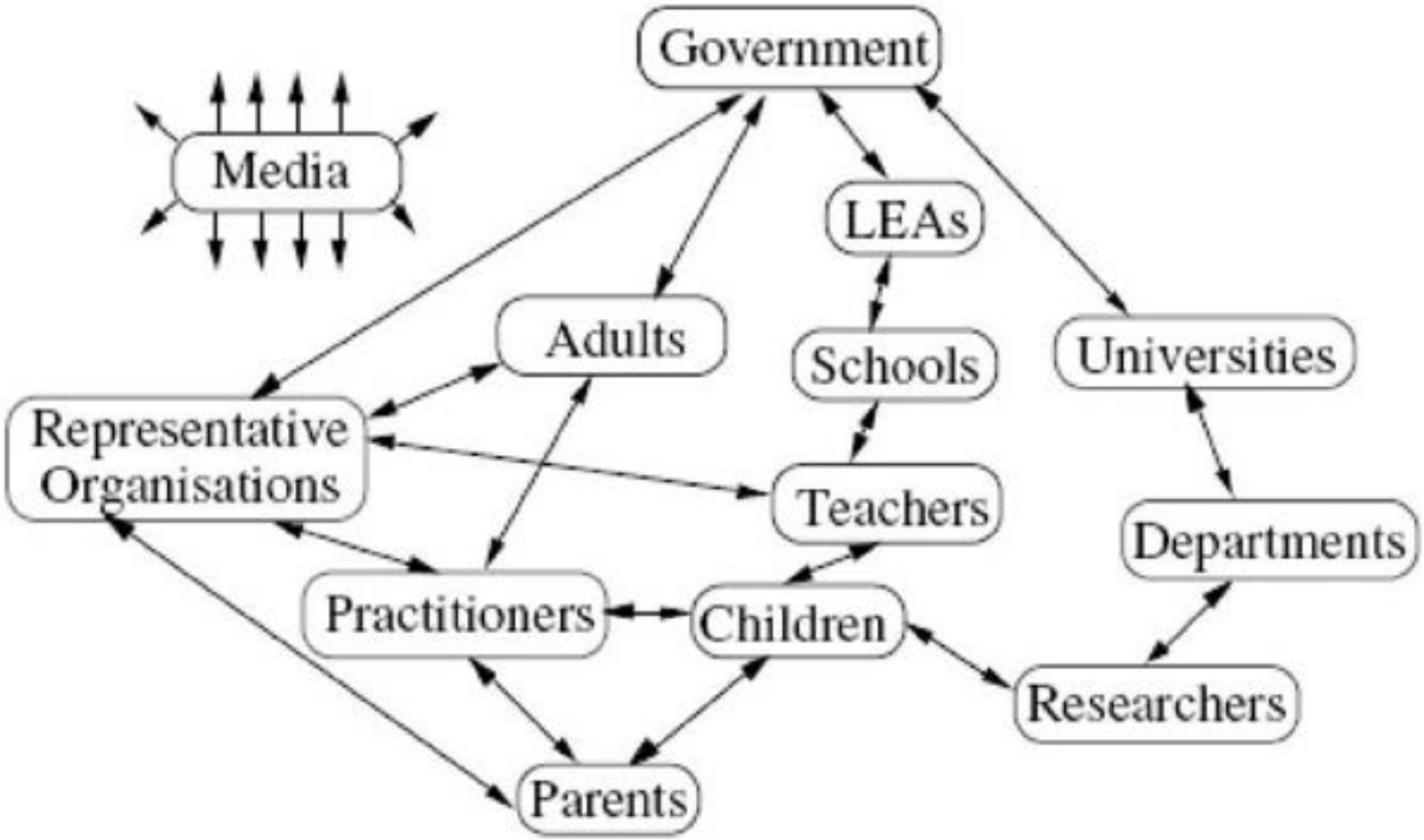
Specific learning disorders

- Neurodevelopmental disorders, typically diagnosed in childhood. Persistent impairment in at least one of three major areas: reading, written expression, and/or math. If not recognized and managed, can cause problems throughout a person's life beyond having lower academic achievement.
- Prevalence: 5 to 15%; 80% of which is in reading (dyslexia). High comorbidity of specific learning disorder with other neurodevelopmental disorders (such as ADHD) as well as anxiety.
- Specific skills that may be affected include word reading accuracy, spelling, grammar, or calculation. In addition, fluency in reading and mathematics may be noted.
- Terminology: Specific learning disorder is a medical term. Learning disability is not exactly synonymous with specific learning disorder but a diagnosis of specific learning disorder can expect to meet criteria for a learning disability and have the legal status of a federally recognized disability to qualify for accommodations and services in school.

"Learning difference" is a term that has gained popularity, especially when speaking with children about their difficulties, as it does not label them as "disordered.

(APA, Physician Review, 'What is specific learning disorder?', 2021)

The Dyslexia Ecosystem



Nicolson, 2002

Assessment: identifying characteristic features of disability at the individual level

- children who may benefit from interventions
- individual areas of strength and weakness

Taxonomy: grouping cases according to distinguishing characteristics.

- classification with implications for underlying aetiological mechanisms
- phenotypes, co-occurrence, developmental trajectories

Diagnostic Criteria: Specific Learning Disorder (DSM-V)

A. Difficulties learning and using academic skills, as indicated by the presence of [symptoms, e.g., reading] that have persisted for at least 6 months, despite the provision of interventions that target those difficulties.

B. The affected academic skills are substantially below those expected for chronological age, and interfere with academic or occupational performance, as confirmed by individually administered standardised achievement measures

C. The learning difficulties begin during school age years, but may not manifest until the demands for those affected academic skills exceed the individual's capacities

D. The learning difficulties are not better accounted for by intellectual disabilities, uncorrected [sensory] acuity, other mental or neurological disorder, psychosocial adversity, lack of proficiency [in language of instruction], inadequate educational instruction.

Dyslexia is an alternative term for a pattern of difficulties characterised by problems with accurate or fluent word recognition, poor decoding, and poor spelling abilities.

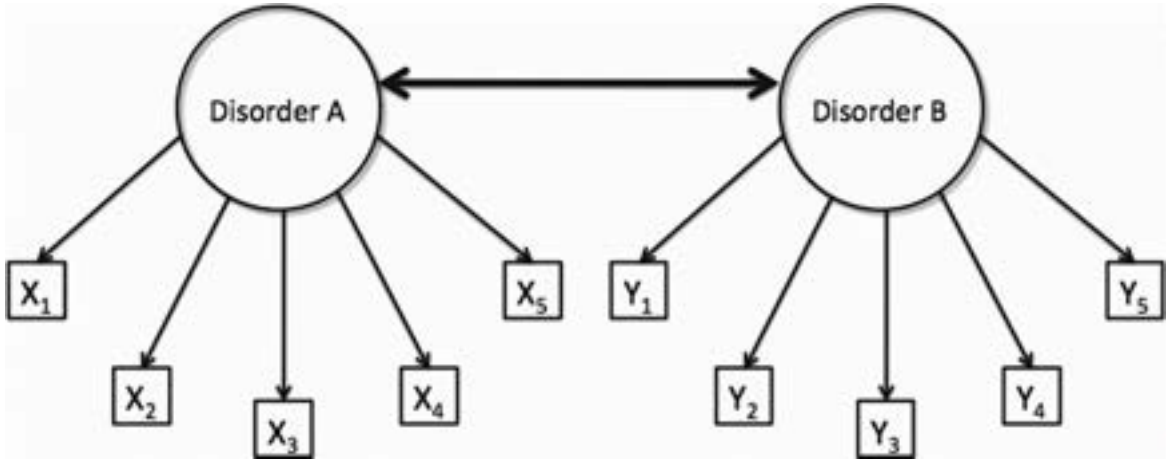
APA (2013)

The Rose Review (2009) Definition

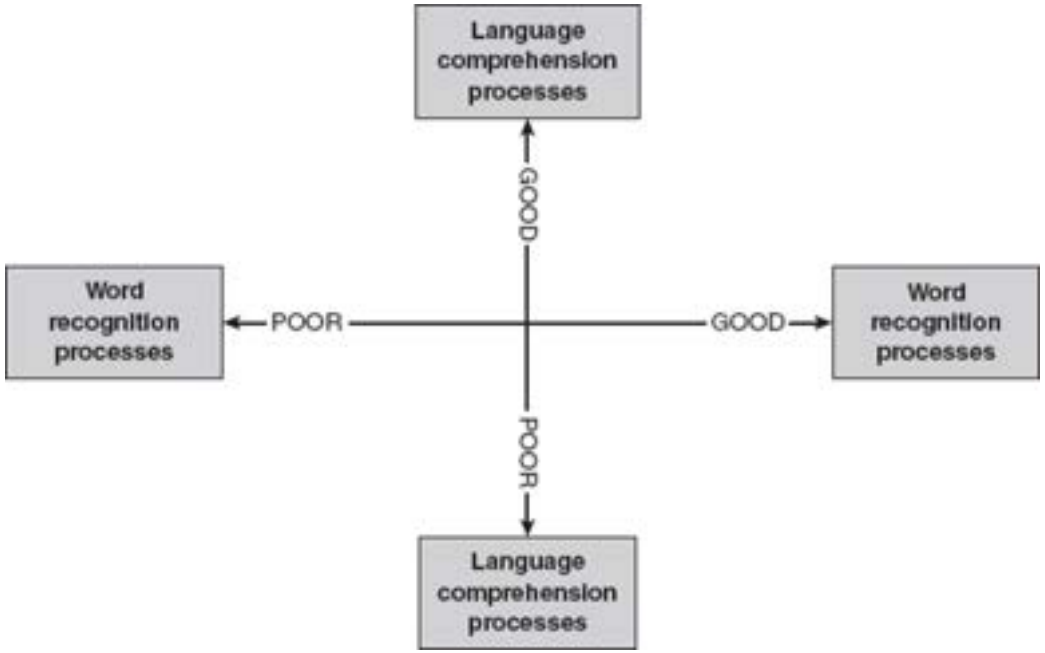
- Educational definition, based on functional deficits in academic achievement
- Alternative approach to use of exclusionary criteria and categorical diagnosis.
- Dyslexia :
  - learning difficulty that primarily affects skills involved in accurate and fluent word reading and spelling.
  - is characterised by difficulties in phonological awareness, verbal memory and verbal processing speed.
  - occurs across the range of intellectual abilities.
  - is best thought of as a **continuum**, rather than a distinct category, with no clear cut-off points.
- **Co-occurring difficulties** , for e.g., in language, motor coordination, concentration, are not independent markers of dyslexia.

Alternative approaches to disorder taxonomies

Categorical



Dimensional



Same symptoms, different disorders?

- Children typically have normal [sensory skills] and intelligence. However, they have also been observed to....

  - ✓ have trouble paying attention to and remembering information presented orally
  - ✓ have problems carrying out multistep directions
  - ✓ have poor listening skills
  - ✓ need more time to process information
  - ✓ have low academic performance
  - ✓ have behaviour problems
  - ✓ have language difficulty
  - ✓ have difficulty with reading, comprehension, spelling, and vocabulary

- ADD/ADHD?
- Communication disorder?
- Dyslexia?

(Auditory Processing Disorder)

NIH Pub. No. 01-4949

A dimensional view of reading

Simple view of reading

(Gough and Tunmer, 1986)

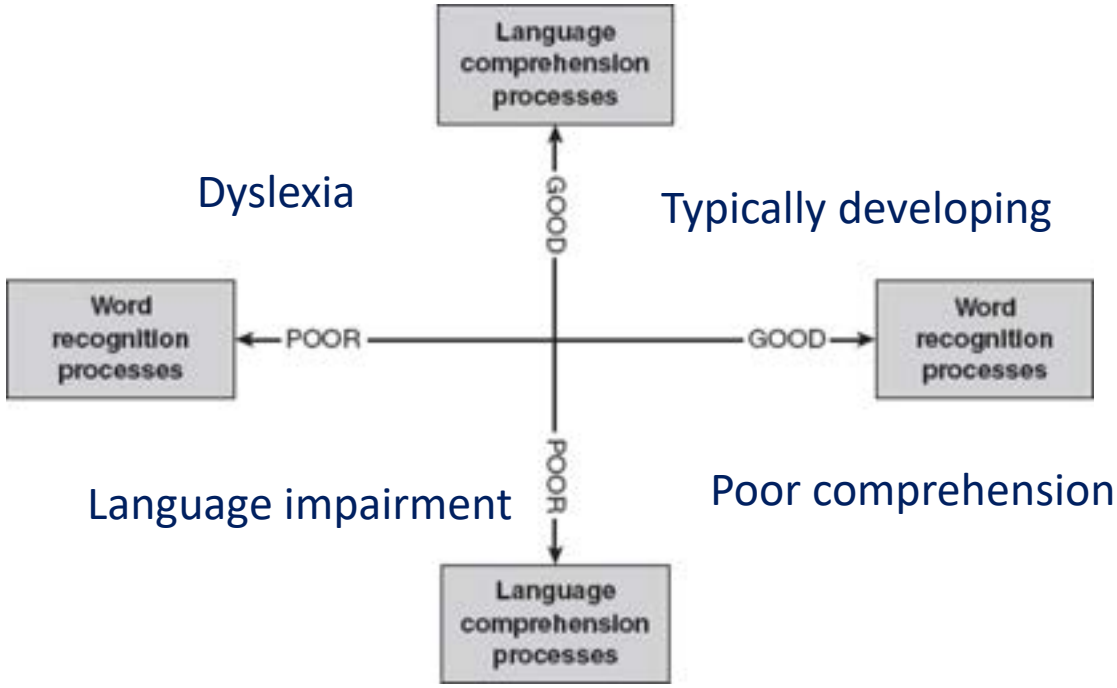
$$RC = WD \times LC$$

RC: reading comprehension

WD: word-level decoding

LC: language comprehension

- Separate but interacting constraints on reading comprehension:
  - learning to read words accurately and fluently
  - having the vocabulary, knowledge, and reasoning skills to support language comprehension more generally

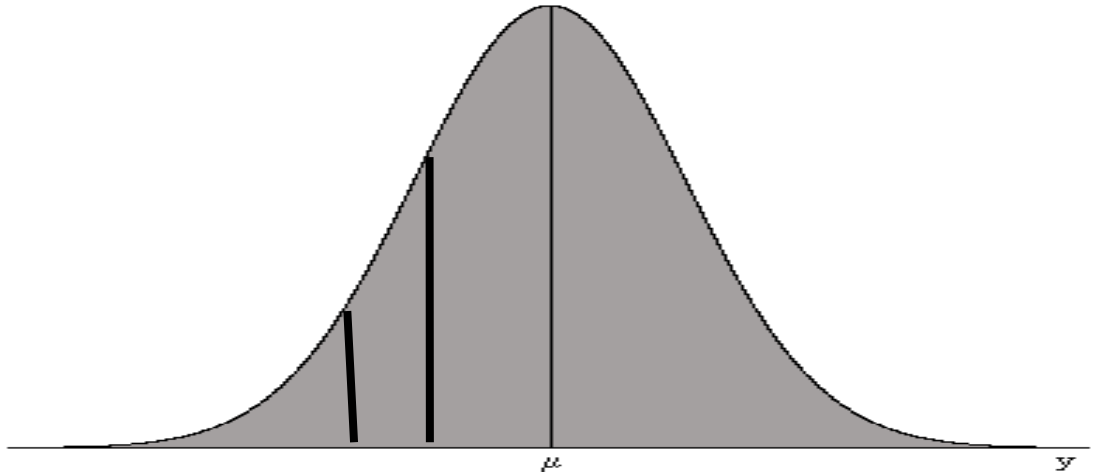


Bishop & Snowling, 2004

Who is ‘dyslexic’?

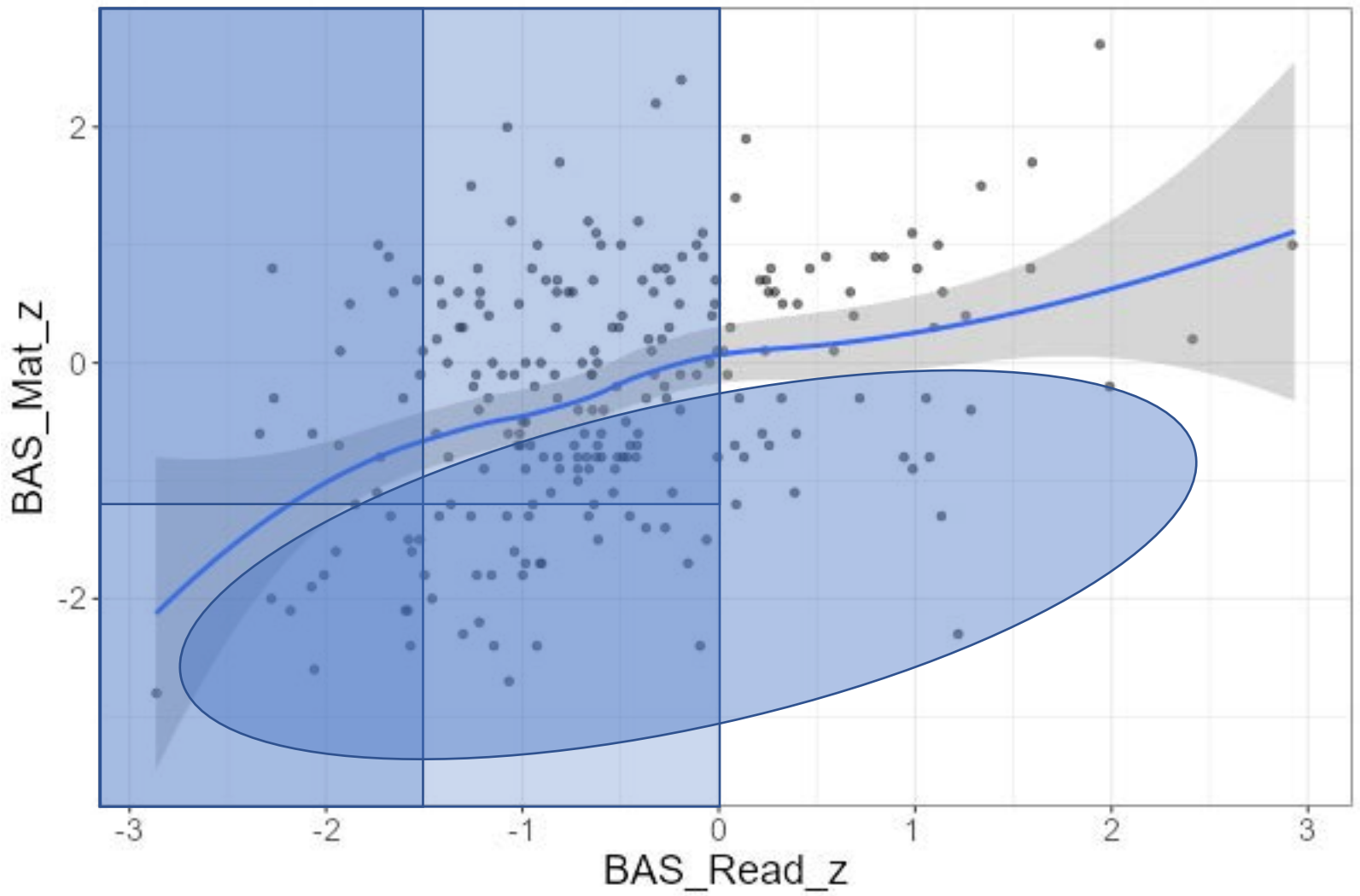
Diagnostic assessment typically depends on:

- Assessment at behavioural level
  - Literacy achievement
    - Single word reading
    - Spelling
    - Reading comprehension
  - Dissociations with other ability measures
- A ‘cut-off’ score
  - Statistical definition of ‘impairment’
    - Based on arbitrary divisions in continuous variables
- Exclusionary criteria
  - Ruling out other potential exogenous and endogenous factors
    - Which ones?



Little consensus on inclusionary criteria!

Who is ‘dyslexic’?



Challenges for taxonomies based on between-group studies

- Groups recruited with inclusionary criteria for ‘dyslexia’ that differ substantially across studies
  - ‘dyslexia’ as a ‘jingle’ fallacy
    - ‘jingle and jangle’ fallacies (Kelly, 1927)
- Within-group heterogeneity is often overlooked or underappreciated
  - Group differences obscure variability within each group
- Case-control designs obscure cross-disorder homogeneity
  - Focus on differences rather than similarities

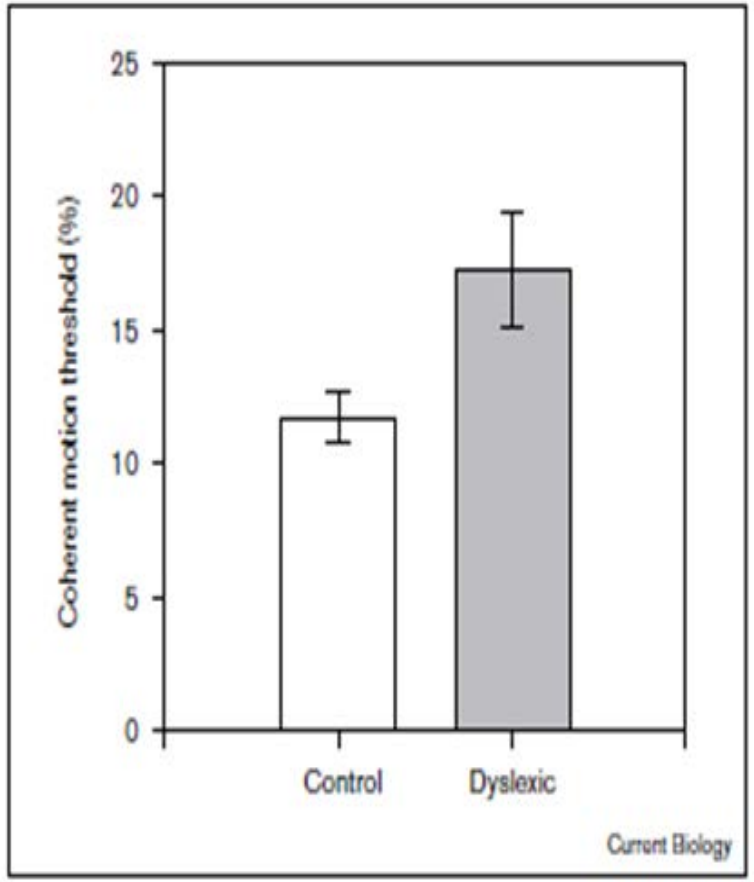


Figure from Witton, Talcott, et al. (1998)

Cognitive and reading dimensions

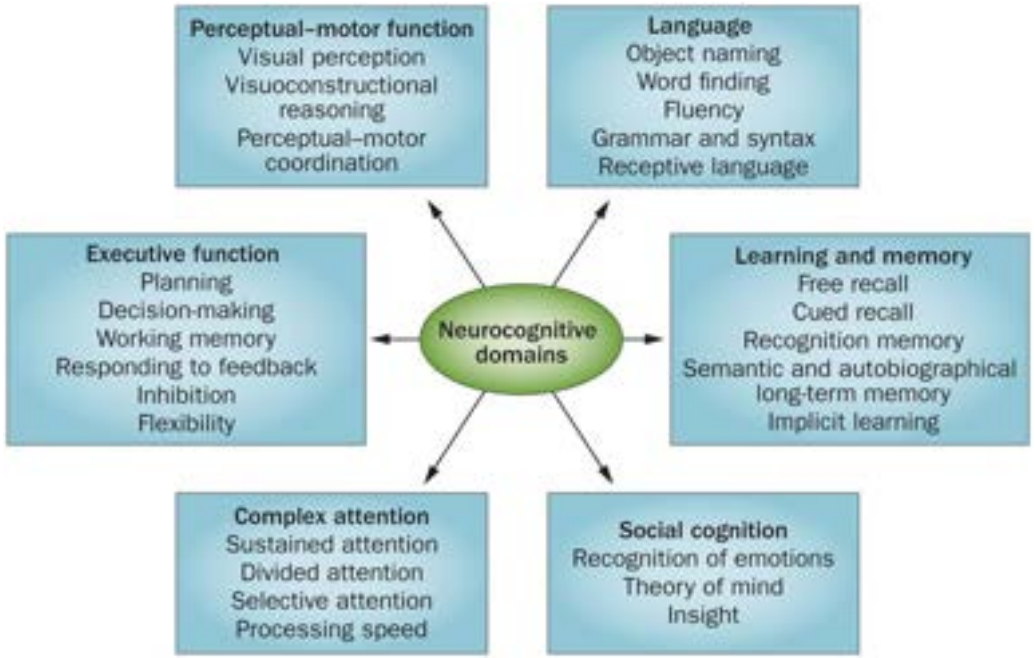
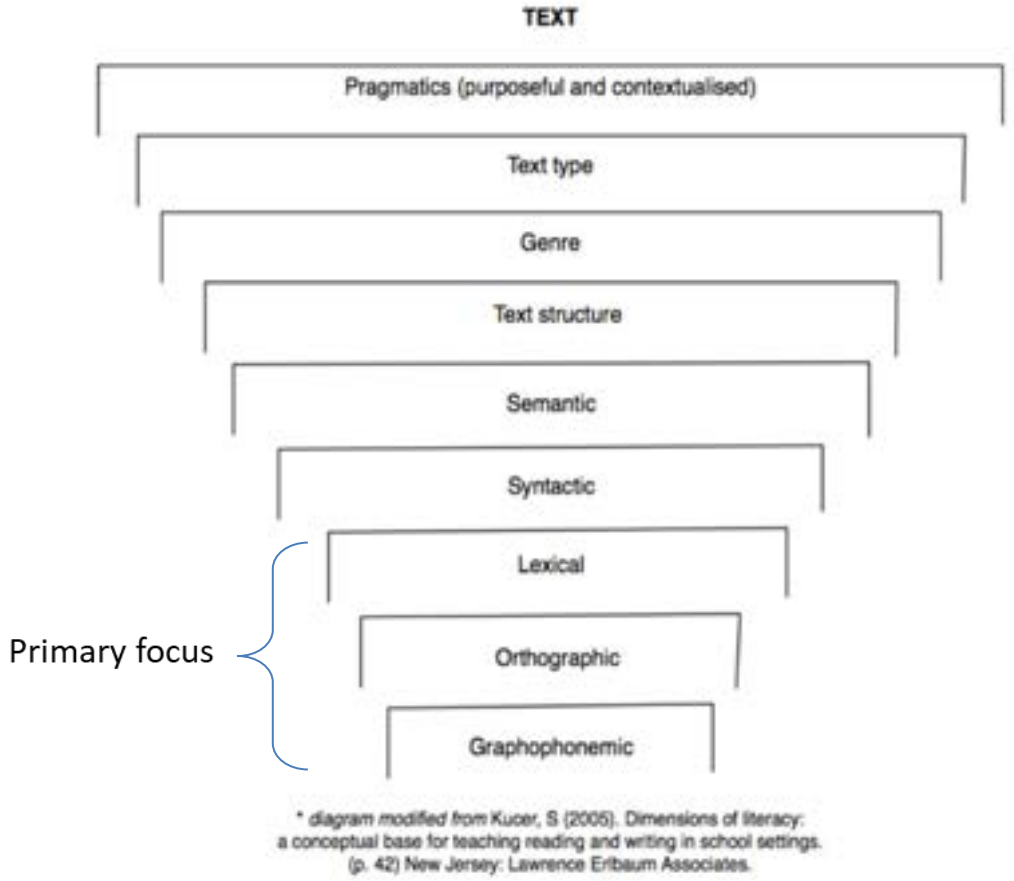


Figure from Sachdev et al, 2014

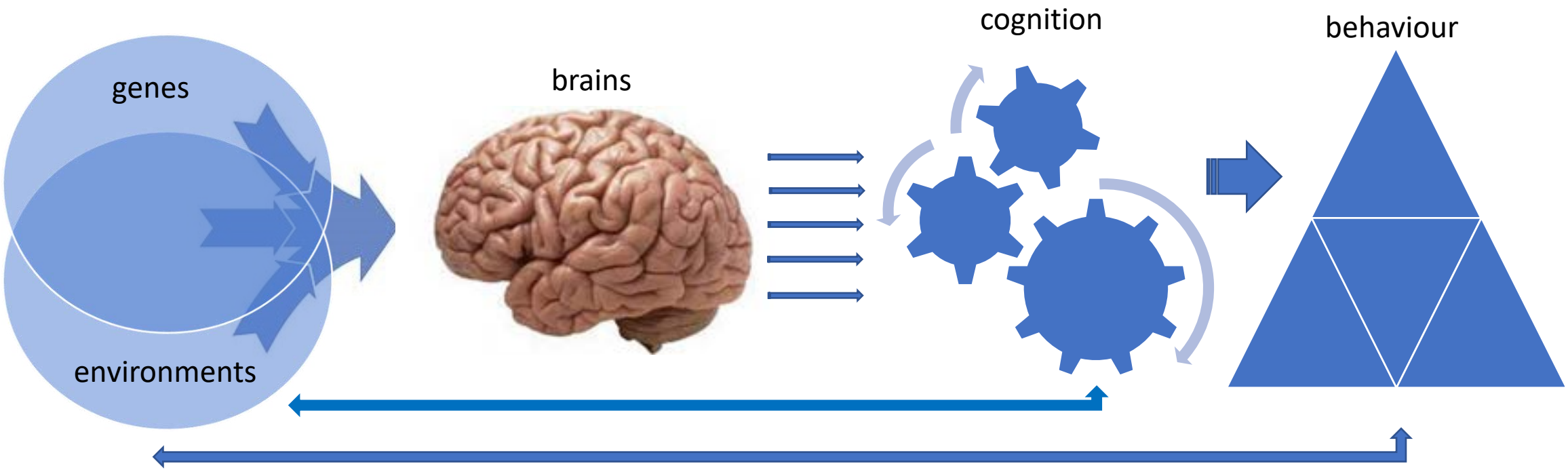


* diagram modified from Kucer, S (2005). Dimensions of literacy: a conceptual base for teaching reading and writing in school settings. (p. 42) New Jersey: Lawrence Erlbaum Associates.

Challenges for diagnostic and assessment practice

1. Pure phenotypes are relatively rare
2. Assessment criteria are typically derived from continuous variables
3. Diagnostic overlap occurs much more frequently than would be expected by chance
4. Diagnoses can be relatively insensitive to the role of development
5. Underlying risk factors act probabilistically not deterministically

Dimensional models of (a)typicality



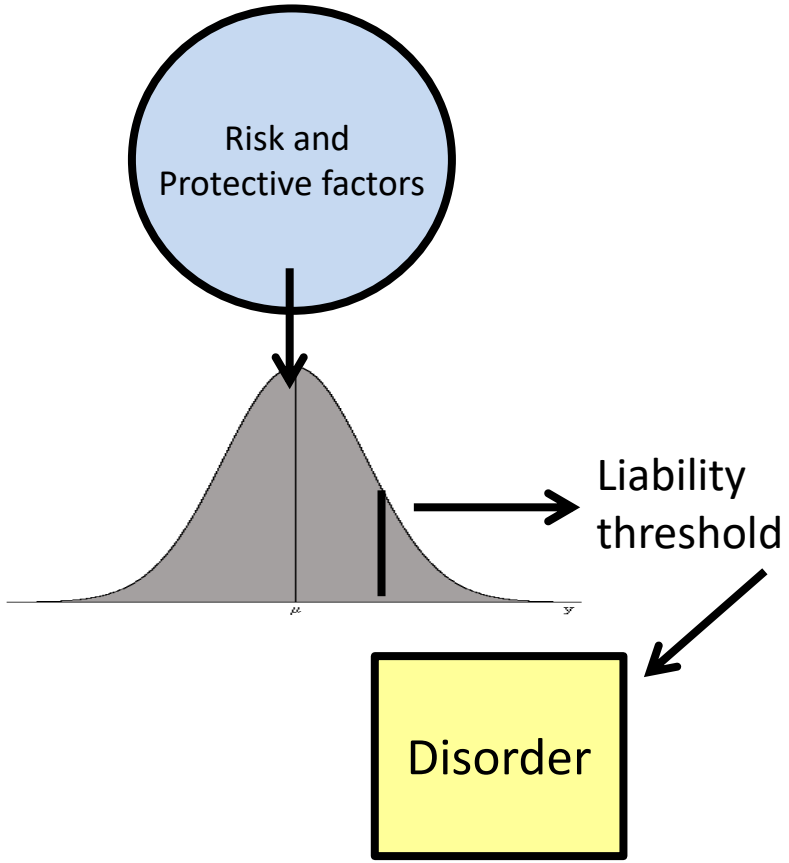
A multilevel framework for conceptualising underlying mechanisms of neurodevelopmental disorders, and of their relationships across different levels of analysis.

Key behavioural dimensions, which currently serve as the primary basis of disorder diagnosis and classification are underpinned by a set of cognitive and biological risk factors, with variable expression and impact across individuals.

Figure 2 from SASA consultation paper from Talcott, 2021

Aetiology in dimensional models

Probabilistic



Liability threshold model: e.g, Neal, 2005

Multidimensional

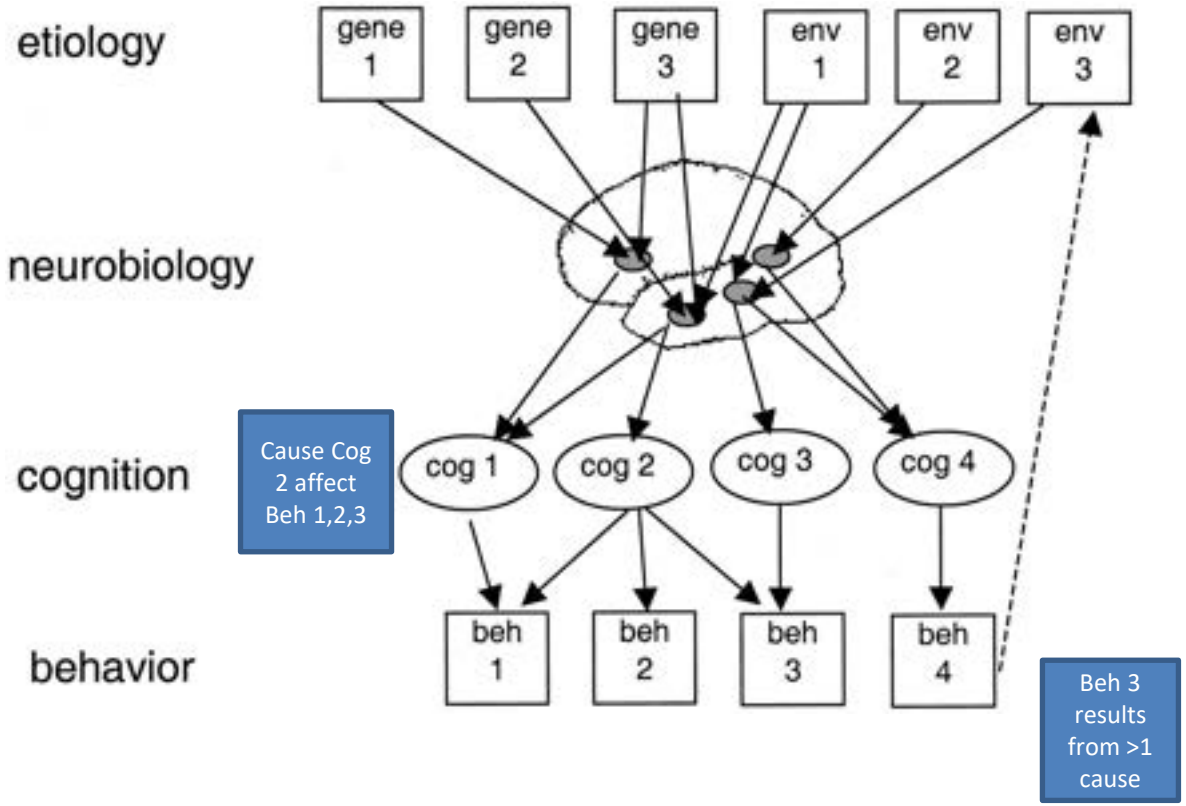


Figure from Bishop and Snowling (2004)

Humans have only been reading and writing for about 5,000 years…

The Phaistos disk (c 1600 BC)

The Phaistos disk (c 1600 BC)

…Unlikely that there is a ‘reading’ or ‘dyslexia’ gene’….

Reading utilizes a **network** of more generic cognitive processes involved with language, perception and thinking.

…and must be learned.

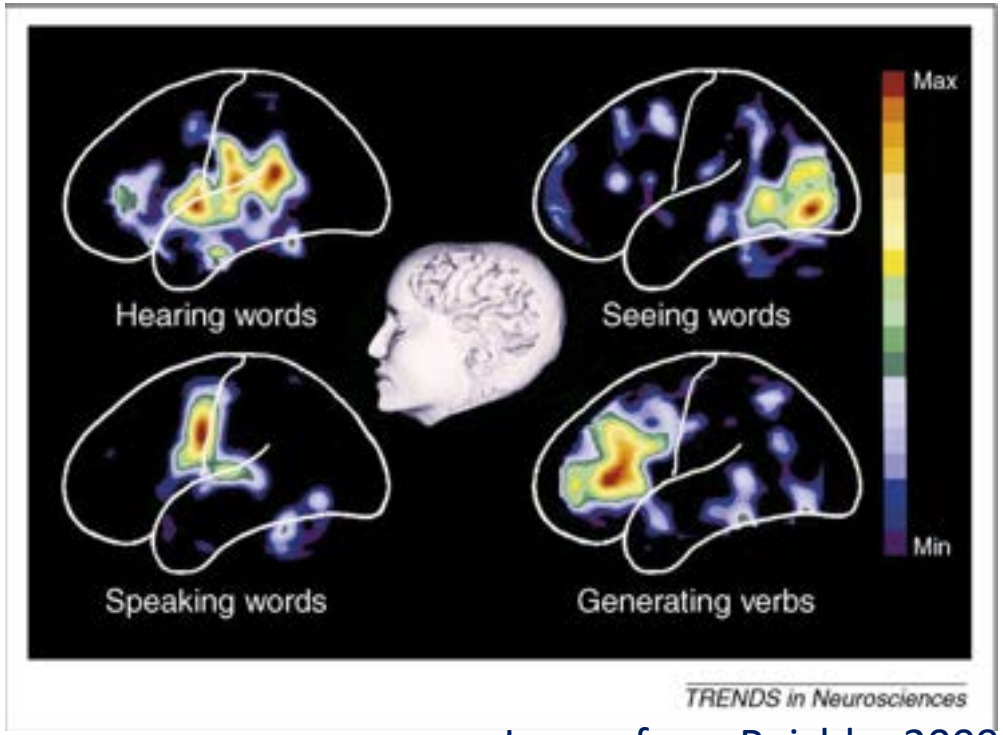
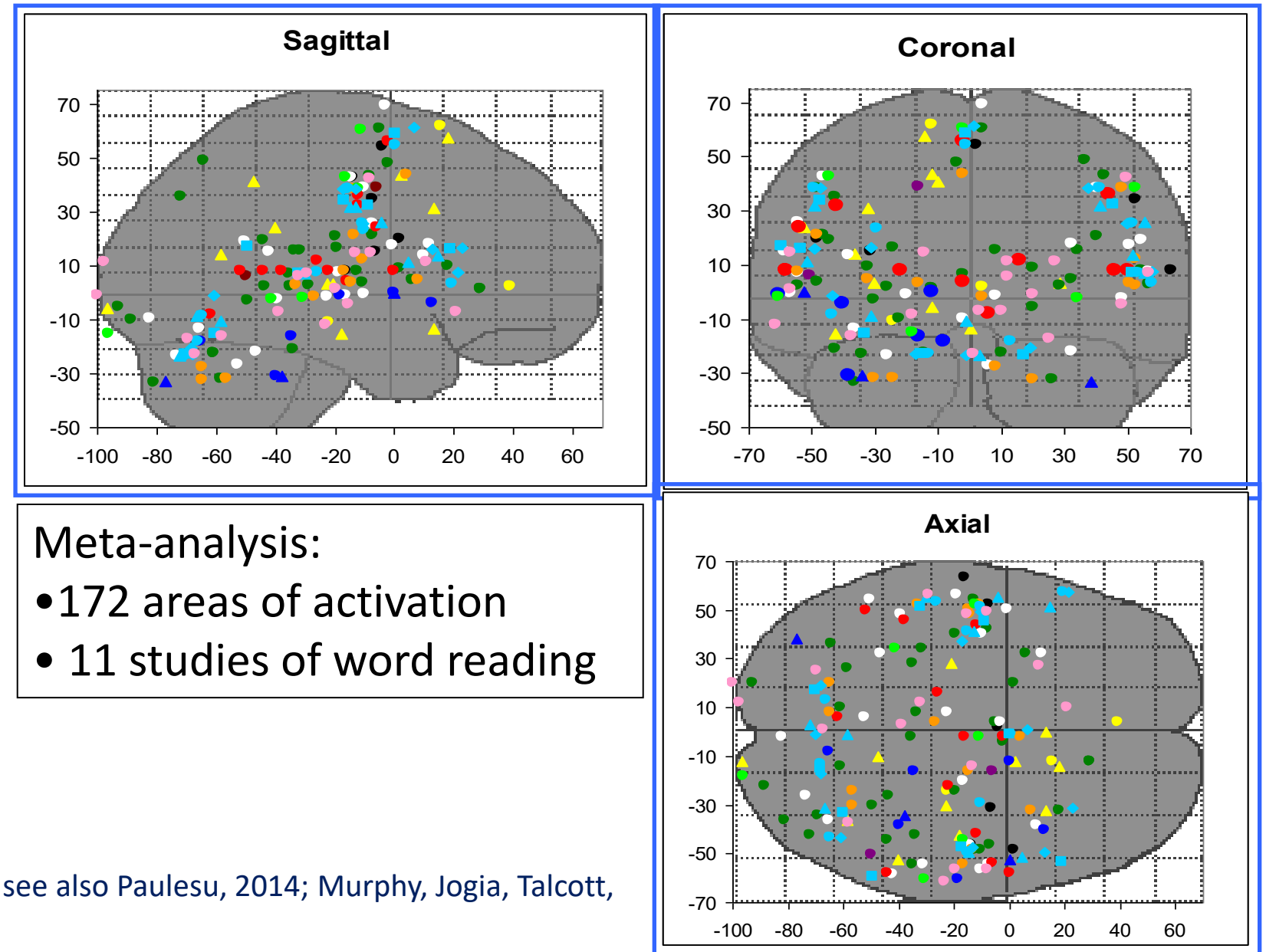


Image from Raichle, 2009

Reading: where in the brain?

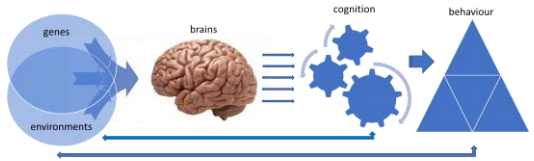


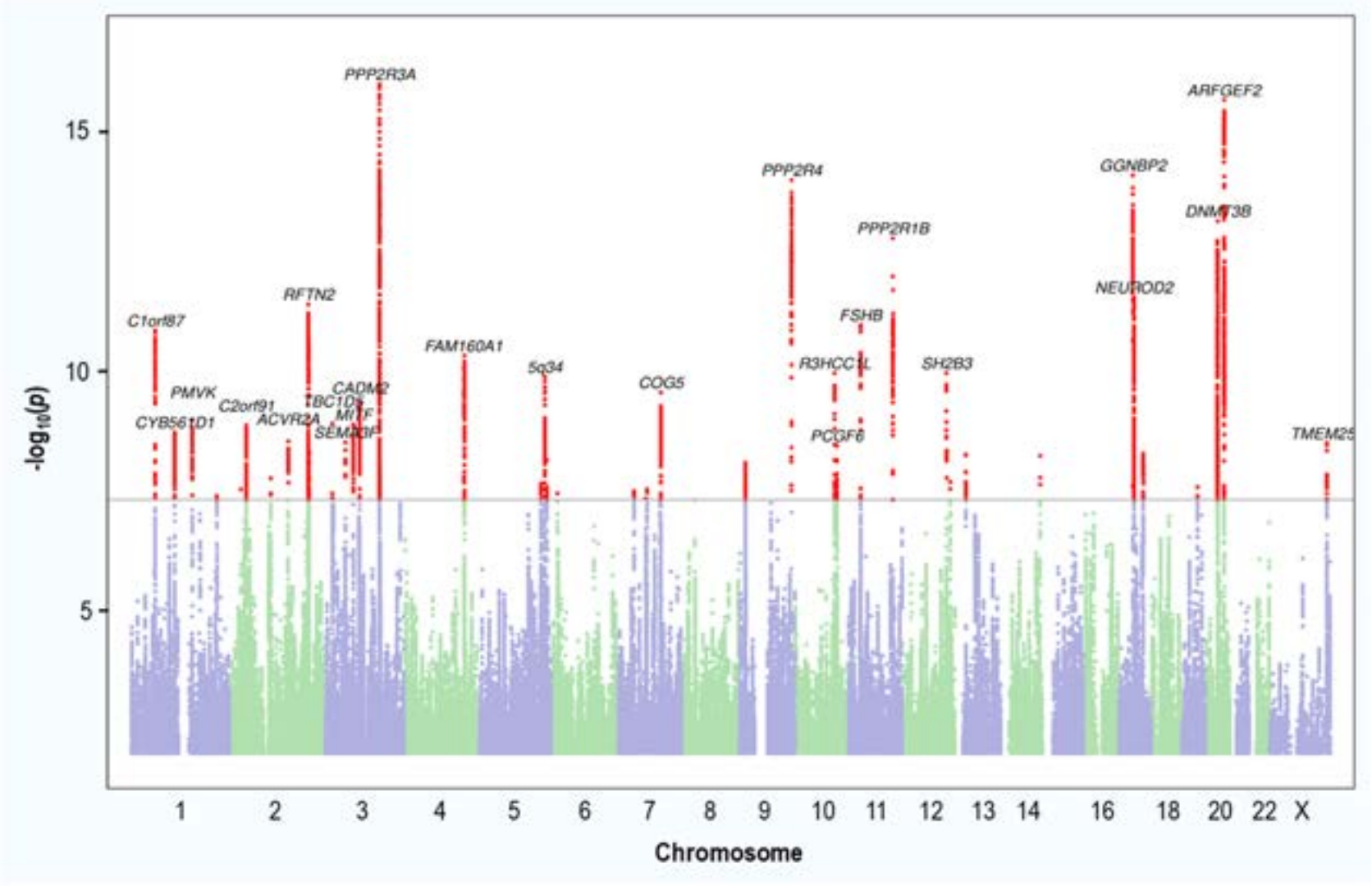
Meta-analysis:

- 172 areas of activation
- 11 studies of word reading

Turkeltaub et al. (2002); see also Paulesu, 2014; Murphy, Jogia, Talcott, 2013

Genes and environments: genetic risks



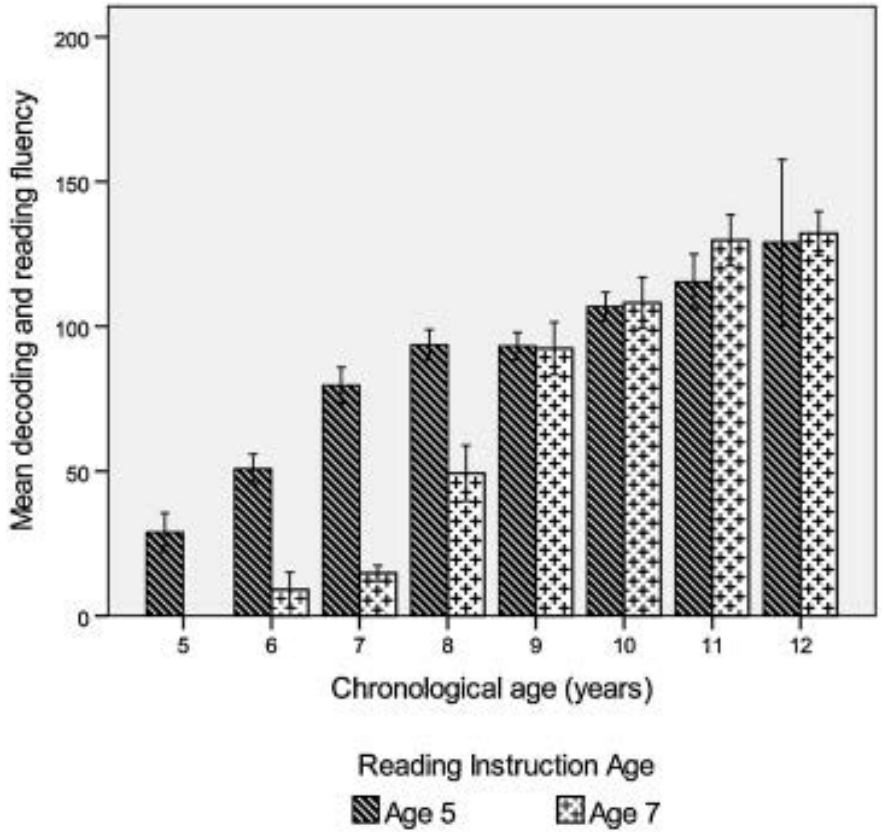


- 23andMe survey
- "Have you been diagnosed with dyslexia?"
  - 51,800 adults self-reporting a dyslexia diagnosis
  - 1,087,070 controls
- 42 independent genetic loci
- 15 linked to cognitive ability/educational attainment;
- 27 novel loci.

(Doust et al., Nature Genetics, in press)

Genes and environments: environmental variables.

Impact of varying age of onset of instruction



Suggate, Schaughency & Reese (2013)

Impact of orthographic transparency (within L1 or between L1 and L2)

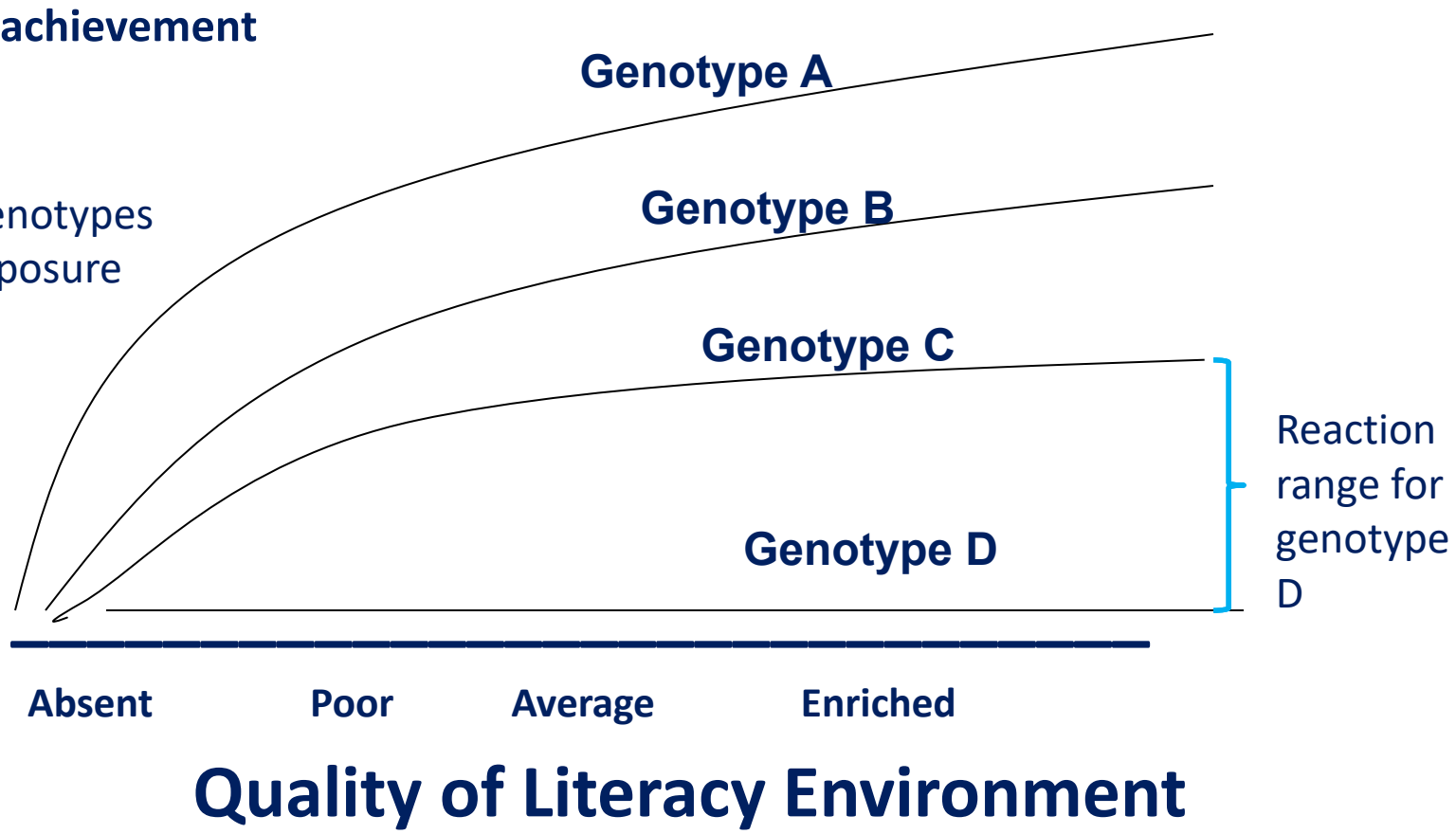
Grapheme-to-phoneme correspondence	Ordering	Language
Transparent/shallow	1	Finnish
1 grapheme-1phoneme	2	Welsh
	3	Italian
	4	Ladin
	5	Serbo-Croatian
	6	Macedonian
	7	Spanish
	8	Catalan
	9	Portuguese
	10	Korean
	11	Hindi
	12	German
	13	Danish
	14	Dutch
	15	Lao
	16	Khmer
	17	French
	18	English
	19	Japanese
Opaque/deep	20	Chinese
1 grapheme-many phonemes	21	Arabic
Many GPC exceptions/irregular words	22	Hebrew

(from Fig. 2.4 in Perfetti & Dunlap, 2008)

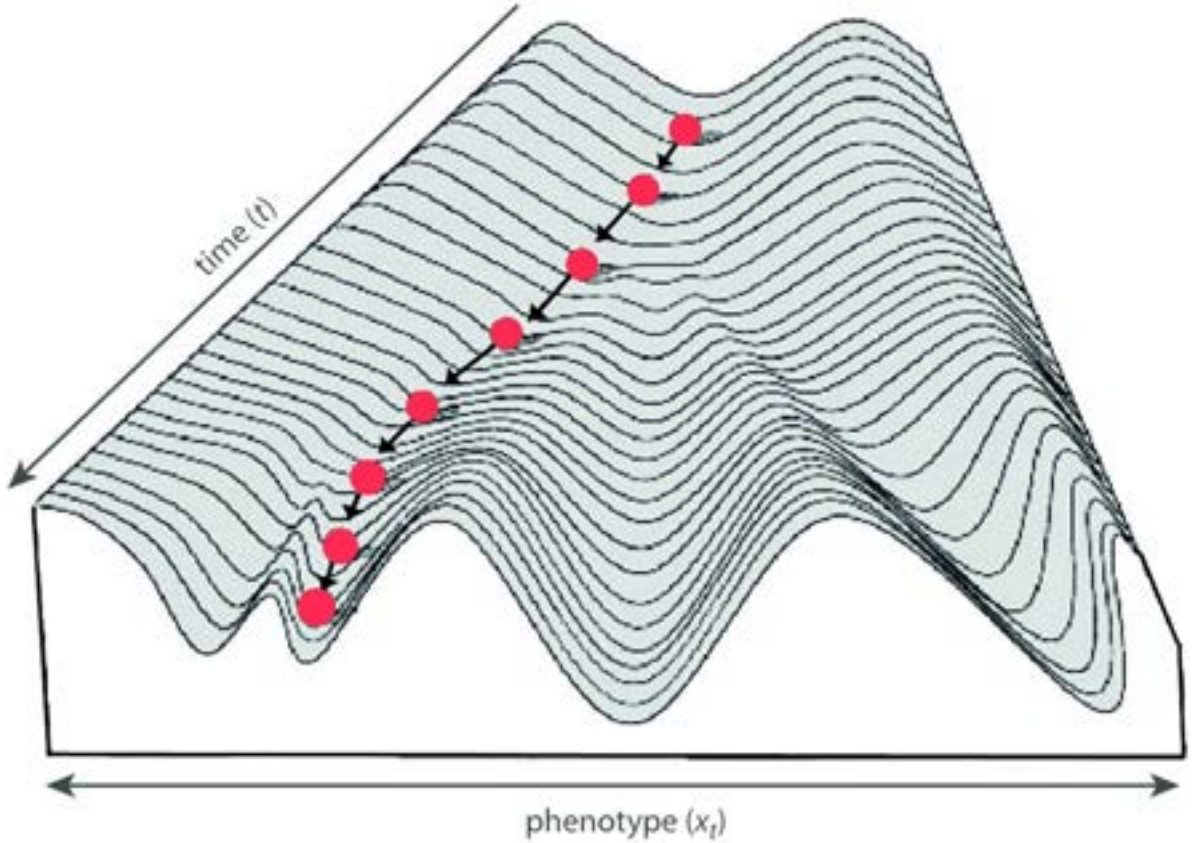
Mechanisms of G x E risks

Range of reaction of different genotypes depending on environmental exposure

- Risk
- Resilience



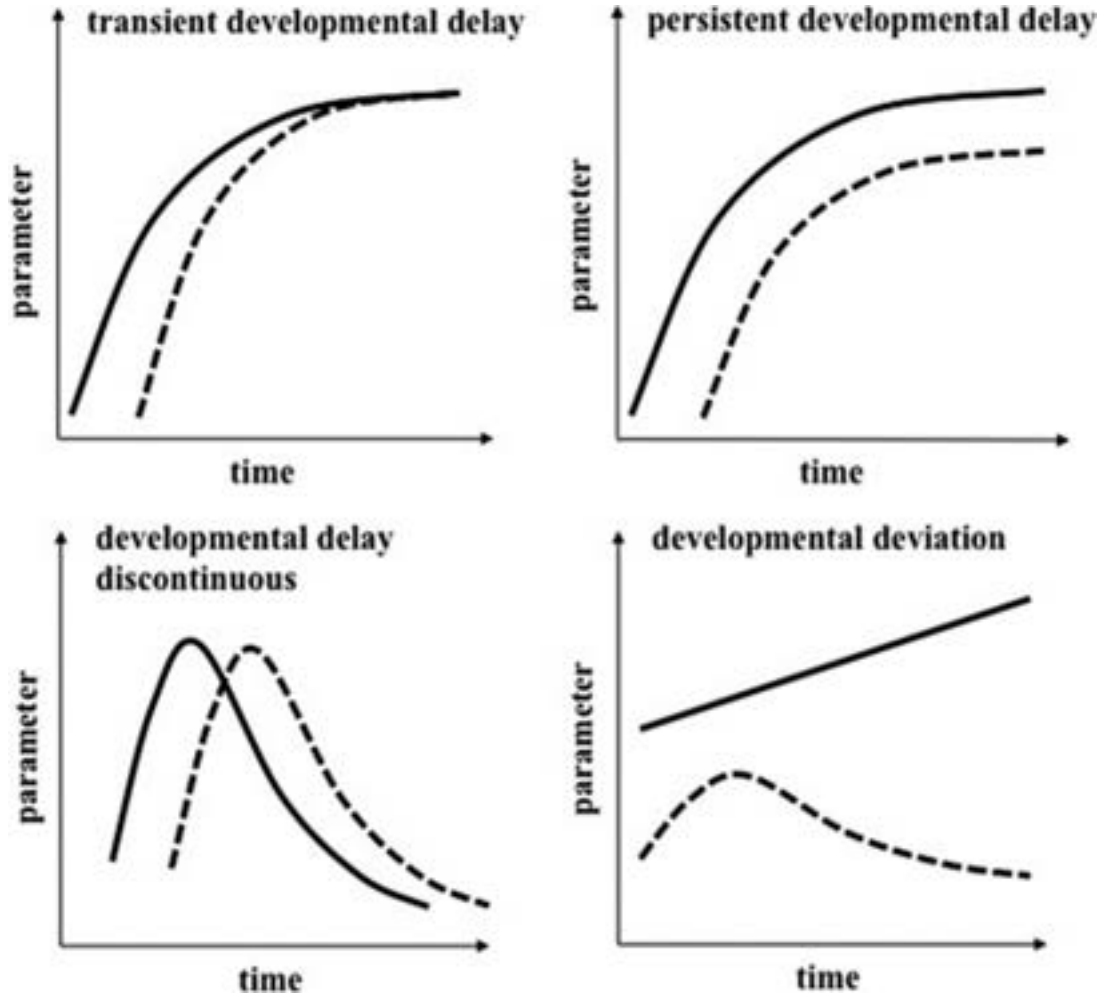
Developmental trajectories



Canalisation

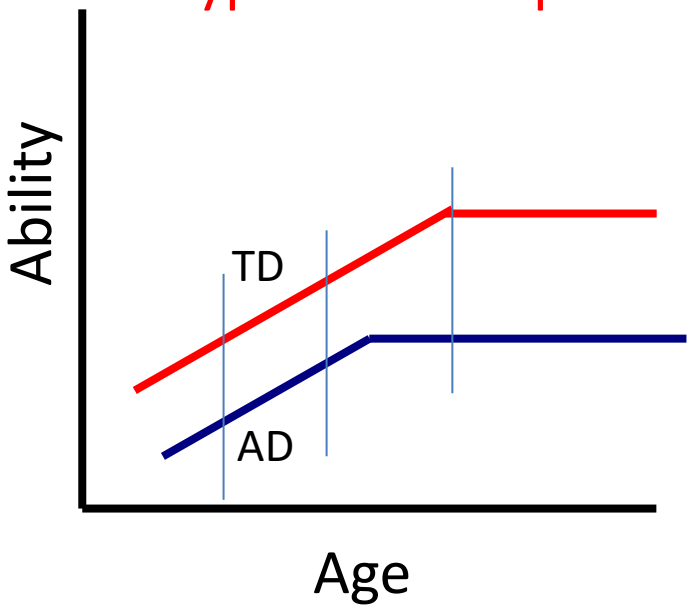
Development restricts number of potential outcomes

More than one pathway, but biology limits the extent to which environments can influence development

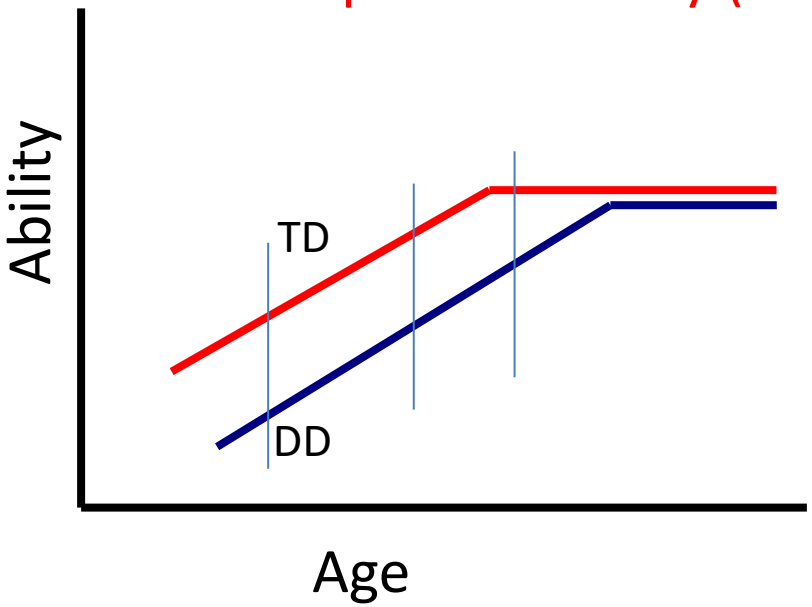


Developmental trajectories particularly relevant to dyslexia screening and assessment

atypical development (AD)



developmental delay (DD)



Persistence

- Sustained underachievement
- Poor response to intervention, effective teaching
- Like not identified from observations made at a single timepoint

Developmental trajectories from longitudinal data

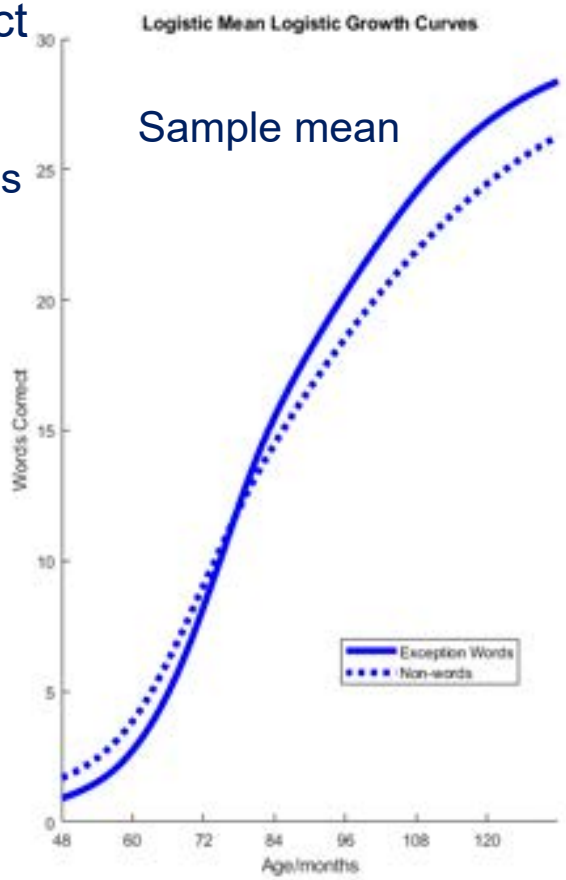
E·S·R·C ECONOMIC & SOCIAL RESEARCH COUNCIL

Aston Literacy Project

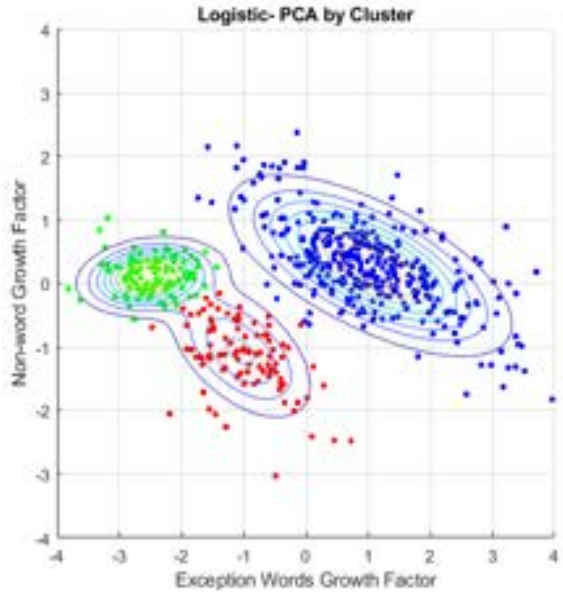
n = 571
- Measured initially as pre-readers

2 parameters for each word type (exception, pseudowords):
- Age to 50%
- Max rate growth

Sample mean



Individual loadings on PCA

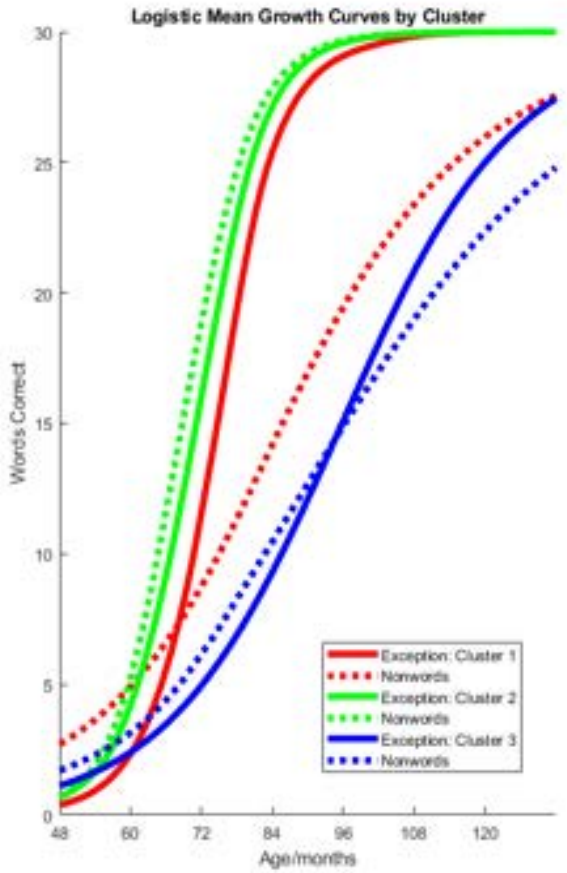


Cluster trajectories

64%

19%

17%



Cunningham, Witton, Talcott, Burgess & Shapiro (2015)
Burgess, Witton, Shapiro & Talcott (2018)

Developmental trajectories of neuro-cognitive development

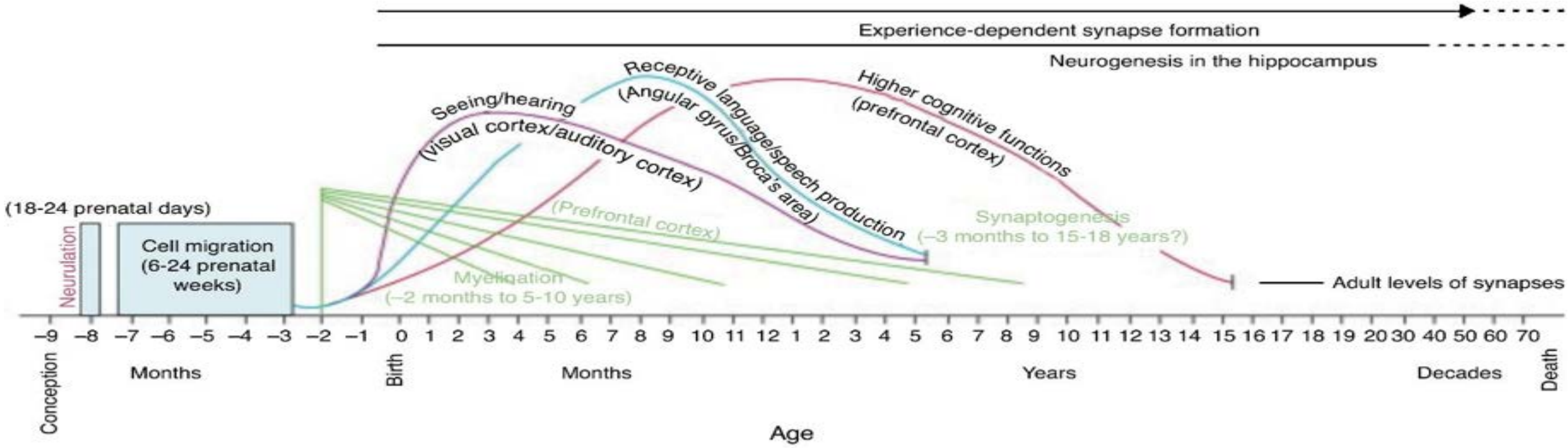
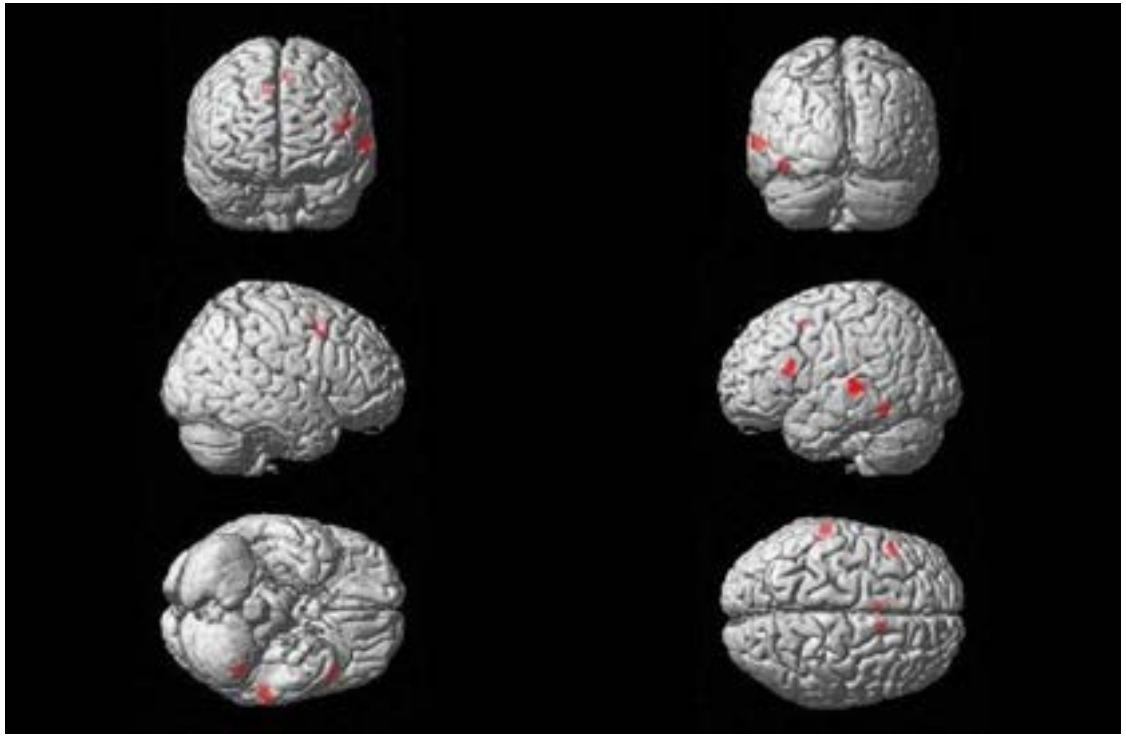
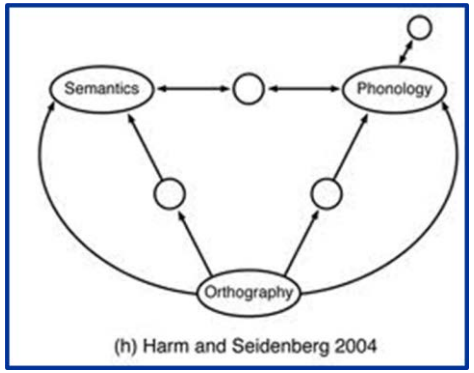


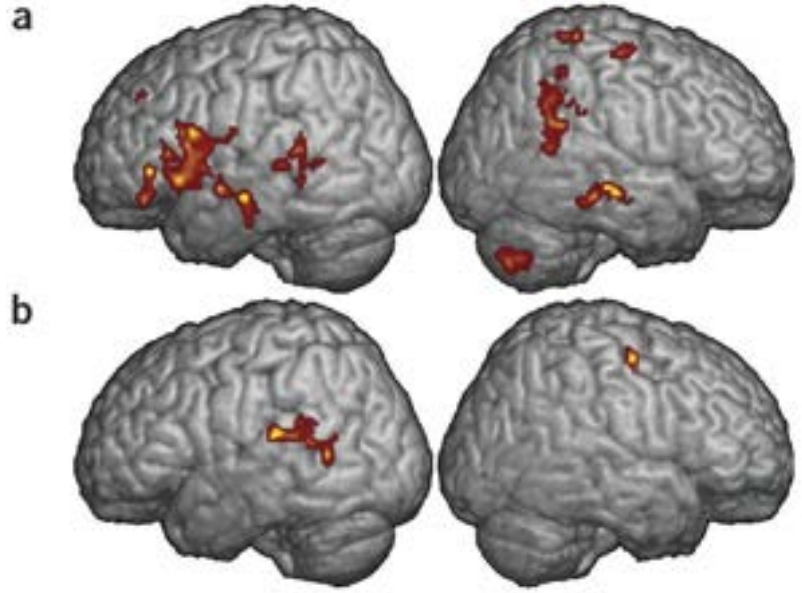
Figure: Thompson & Nelson,2001

The (single-word) reading brain



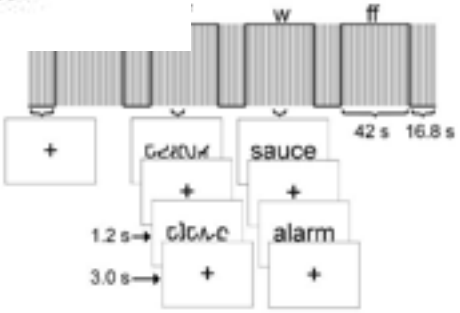


Individual effects



Adults

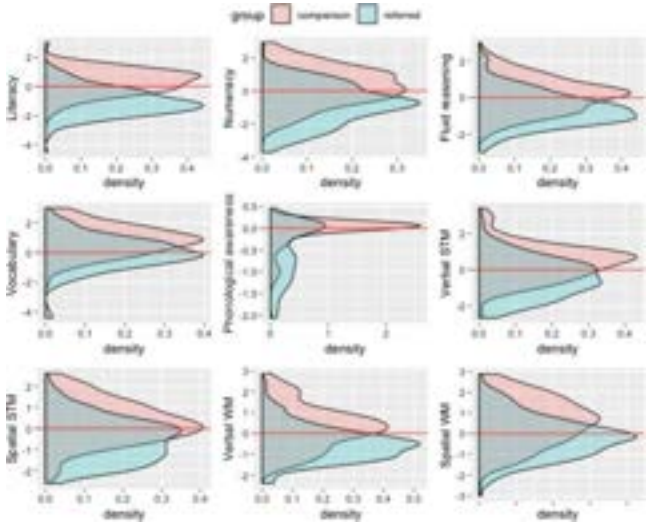
Children



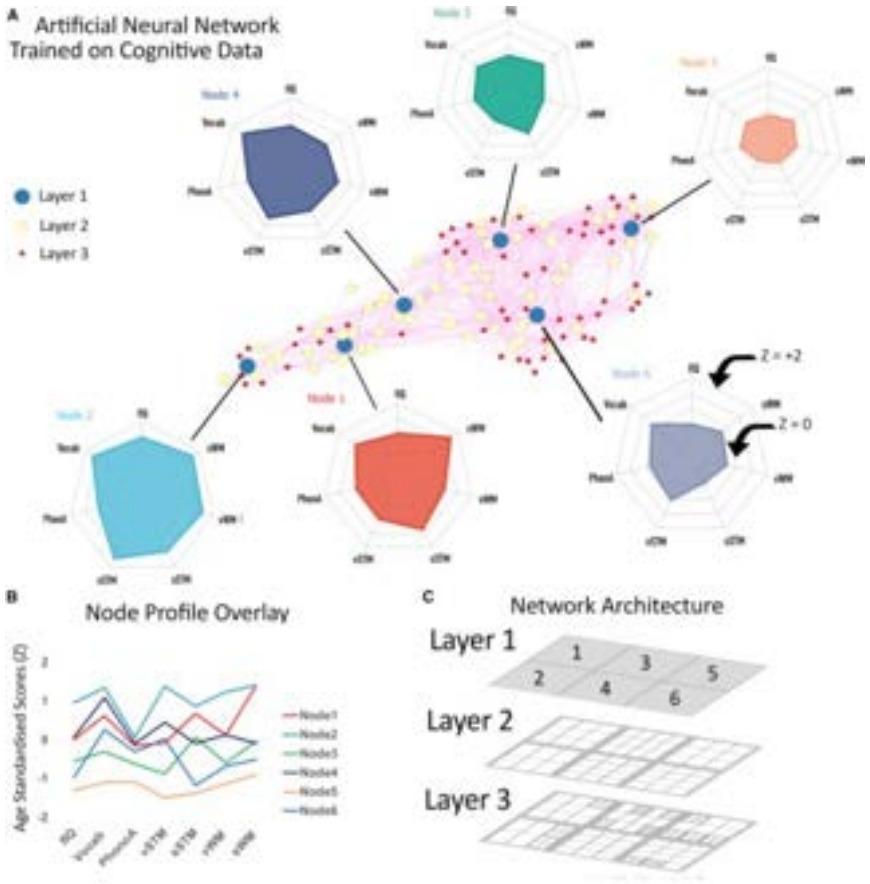
Developmental effects

Turkeltaub et al., 2003

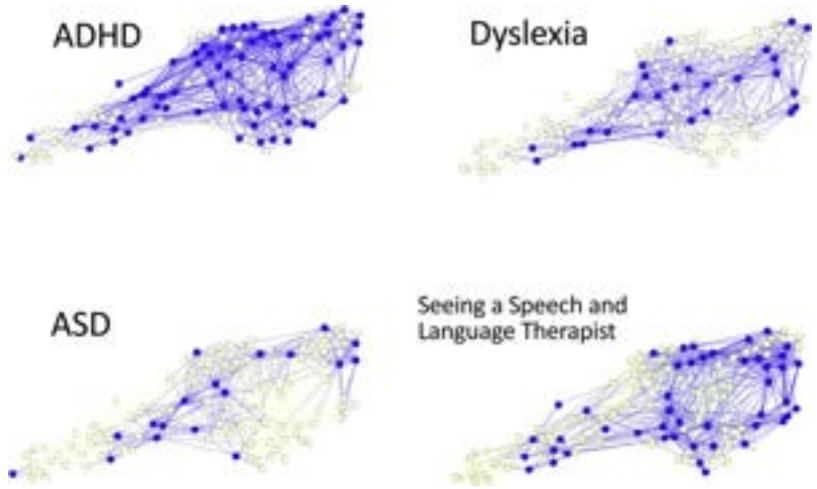
Cluster based approaches



Aggregate Raw data



Multidimensional Cognitive Clusters



Clusters validated against existing diagnostic entities

(Siugzdaite , Bathelt , Holmes & Astle,2020)

Remit of the talk

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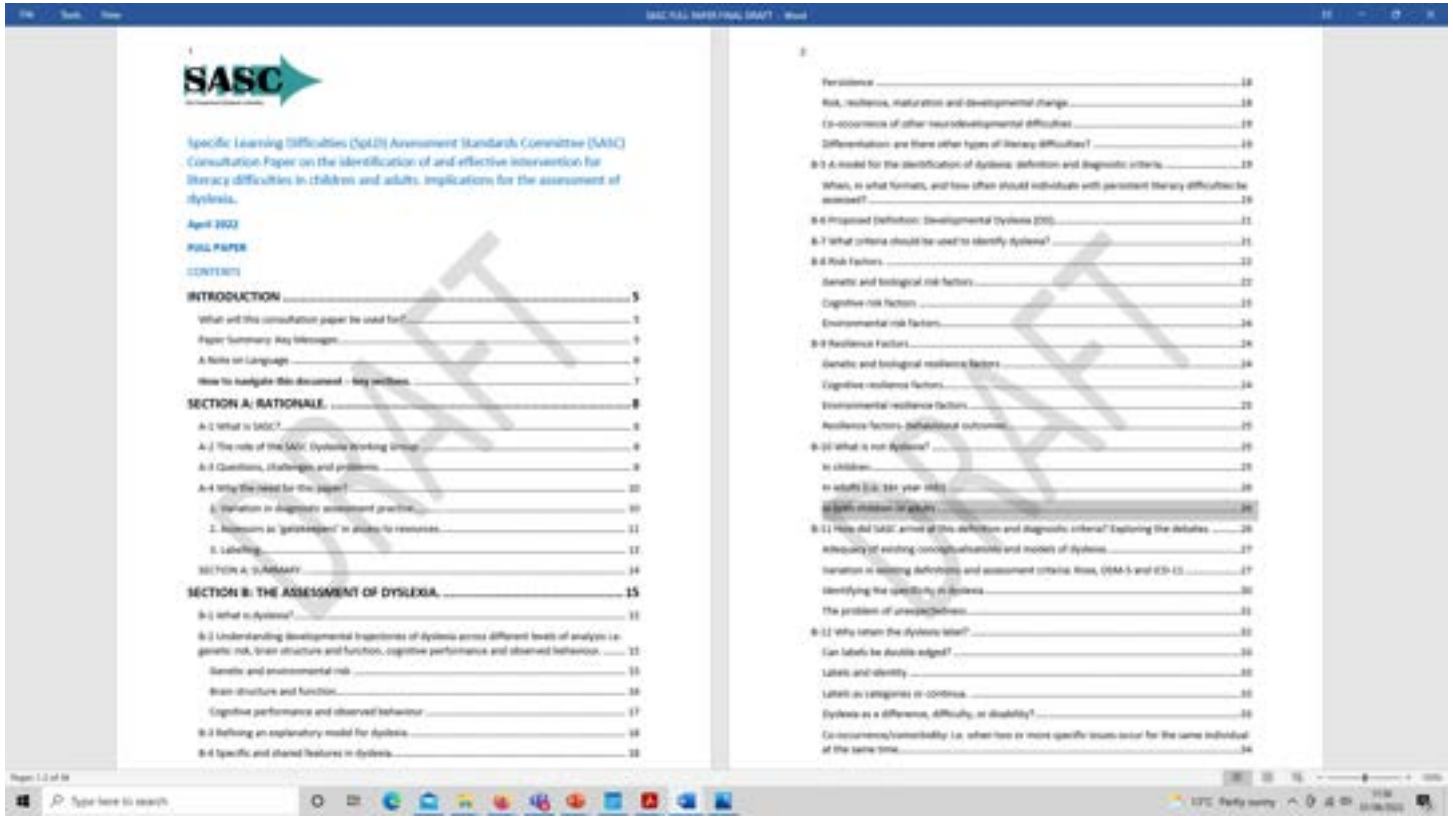

What would be the taxonomic dimensions we would want report?

Proposed Dyslexia taxonomy:

1. Behavioural variation (manifestation, dissociation)
2. Genetic risk (genetic, familial)
3. Organicity (neurological risk)
4. Cognitive risk (attention, memory, language)
5. Development and persistence (trajectory RTI)

NDDs:
Classification, Dimensions and Outcomes

DSMV	INFORMAL LABELS	DIMENSIONS	OUTCOMES
• Specific Learning Disorders (reading, writing, maths • Autism Spectrum Disorder • ADHD • Communication disorders (language disorder, speech sound disorder, social communication disorder)	• Struggling learners (functional) • Poor social skills, "on the spectrum" • Poor attention • Restless • Easily upset, sensitive	• Phonological processing • Language comprehension • Processing speed • Executive control • Pragmatic communication • Inattention • Hyperactivity • Impulsivity/ emotional control • Reward delay tolerance	**Achievement** • Literacy • Maths **Behaviour** • Pragmatic communication • Hyperactivity/ impulsivity • Emotional control • Conduct (aggression)?

Aston University
Birmingham

Thank you!

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