

Dyslexia: a research update

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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.

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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



Taxonomy: grouping cases according to distinguishing characteristics.

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

Assessment: identifying characteristic features of disability at the individual level

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



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.

DSM-5

The Rose Review (2009) Definition

- Educational definition, based on <u>functional deficits</u> 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
 - \checkmark need more time to process information
 - \checkmark 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)

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



Who is 'dyslexic'?

Diagnostic assessment typically depends on:

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





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



Cognitive and reading dimensions



TEXT



Figure from Sachdev et al, 2014

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

Multidimensional



Liability threshold model: e.g, Neal, 2005

Figure from Bishop and Snowling (2004)

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







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...Unlikely that there is a 'reading' or 'dyslexia' gene'....

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

The Phaistos disk (c 1600 BC)

...and must be learned.



Reading: where in the brain?



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.

Genes and environments: environmental variables.

Impact of varying age of onset of instruction



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
4	7	Spanish
	8	Catalan
4	9	Portuguese
	10	Korean
4	11	Hindi
	12	German
4	13	Danish
ſ	14	Dutch
1	15	Lao
	16	Khmer
1	17	French
	18	English
¥	19	Japanese
O pa que/deep	20	Chinese
1 grapheme-many phonemes	21	Arabic
Many GPC exceptions/irregular words	22	Hebrew

Mechanisms of G x E risks



Developmental trajectories



Developmental trajectories particularly relevant to dyslexia screening and assessment



Persistence

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

Developmental trajectories from longitudinal data





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

Developmental trajectories of neuro-cognitive development



The (single-word) reading brain







Individual effects

Turkeltaub et al., 2003

Cluster based approaches



Aggregate Raw data



Multidimensional Cognitive Clusters

(Siugzdaite , Bathelt , Holmes & Astle, 2020)

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What would be the taxonomic dimensions we would want report?

DSM-V Axis

Axis I

clinical disorders and developmental and learning disorders.

Axis 2

- personality disorders or mental retardation, includes autism
- Axis 3
 - medical and/or physical conditions or disorders
- Axis 4
 - Psychosocial & Environmental Problems

Axis 5

 Overall psychological, social, and occupational functioning (scale from 1 – 100)

Proposed Dyslexia taxonomy:

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

NDDs:

Classification, Dimensions and Outcomes

	DSMV	I	NFORMAL LABELS		DIMENSIONS		OUTCOMES
• S C w	pecific Learning Disorders (reading, vriting, maths	•	Struggling learners (functional) Poor social skills.	•	Phonological processing Language comprehension	•	Achievement Literacy Maths
• A C	outism Spectrum Disorder		"on the spectrum"	•	Processing speed Executive	•	Behaviour Pragmatic
• Д	NDHD	•	Restless	•	control Pragmatic	•	Hyperactivity/ impulsivity
 C d s c d 	Communication lisorders (language lisorder, speech ound disorder, ocial ommunication lisorder)	•	Easily upset, sensitive	•	communication Inattention Hyperactivity Impulsivity/ emotional control Reward delay tolerance	•	Emotional control Conduct (aggression)?



Thank you!

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