Risk Factors for Dyslexia? Evidence from Family Risk Studies

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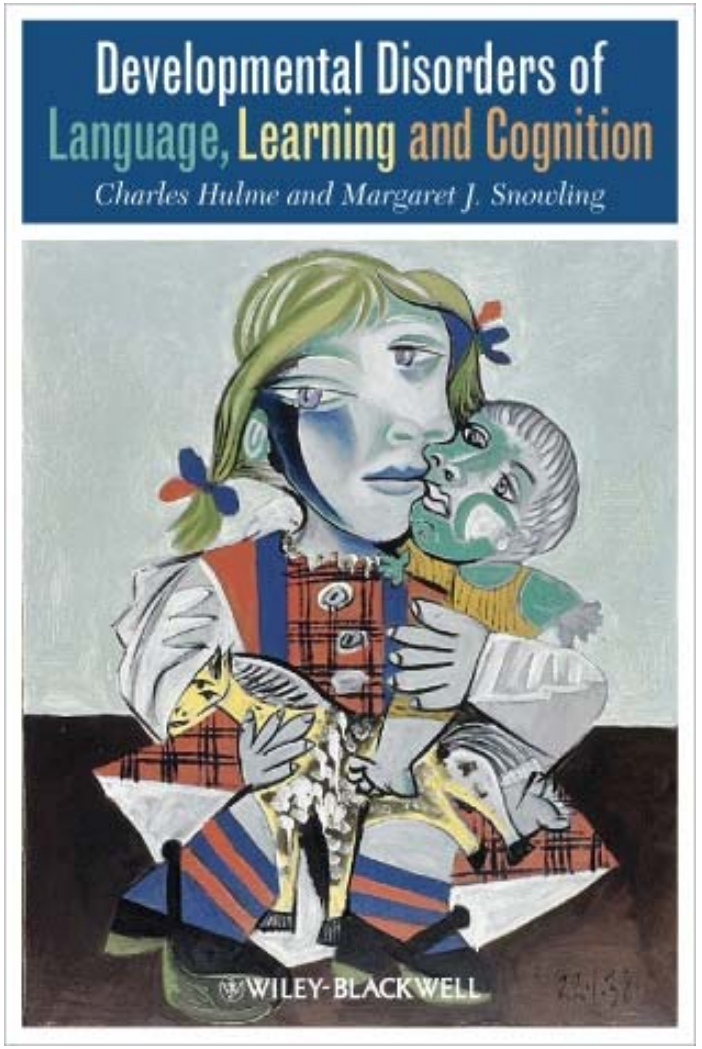
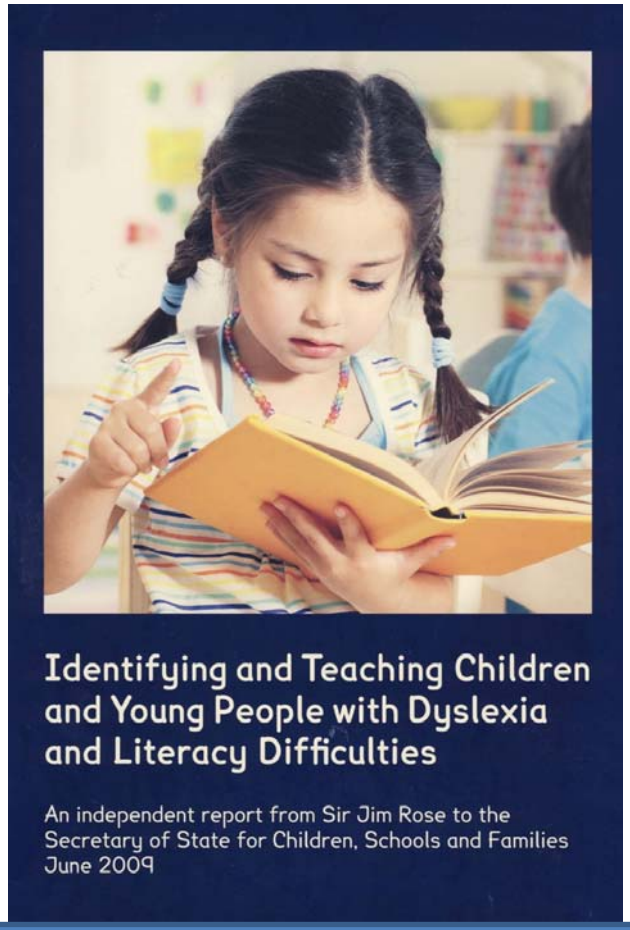
A Developmental Perspective

- *Genetic and environmental factors represent the "ultimate" causes of disorders (like dyslexia).*
- *These causes are probabilistic (risks)*
- *Risk factors are continuous (weaker or stronger)*
- *A genetic risk will be moderated by environmental factors.*
- *Sets of risk factors may interact to give multiplicative (synergistic) effects*



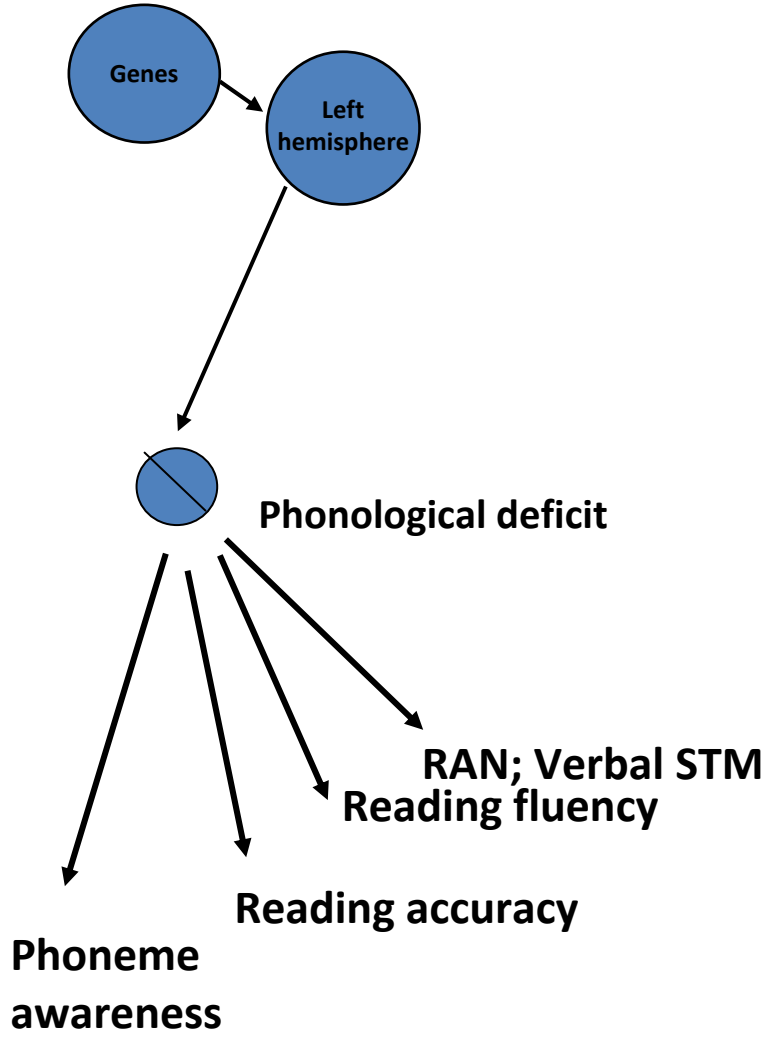
A Dimensional View of Dyslexia



Rose Review 2009
Department for Children Family and Schools

- *Dyslexia occurs across the range of intellectual abilities*
- *It is best thought of as a continuum, not a distinct category, and there are no clear cut-off points.*
- *Co-occurring difficulties may be seen in aspects of language, motor co-ordination, mental calculation, concentration and personal organisation, but these are not, by themselves, markers of dyslexia*

Changing Concepts of Dyslexia



- Dyslexia as a dimension (mild to severe)
- Risk Factors accumulate toward a threshold for identification ('dyslexia' more likely when more than one deficit present)
- 'Diagnosis' depends on agreed external criteria

Take Home Messages

- Longitudinal studies of children at family-risk of dyslexia show that there are two primary risk factors for dyslexia: poor Phonology and poor Language
- Many parents who self-report as dyslexic also have language difficulties and problems of attentional control (which they pass on to their offspring)
- Two patterns of difficulty are observed among pre-school children at family risk of dyslexia: a Language impairment or a specific difficulty with Phonology observed in phonological memory/sentence repetition/verb inflection
- Among older siblings of FR children, those with dyslexia tend to have poor Phonology and poor Language; poor phonology also characterizes unaffected sibs
- Aspects of the home literacy environment may be beneficial to the literacy development of children at family risk

Implication: a phonological deficit is only one cause of dyslexia; language impairments also require attention

What is a Family–Risk Study?

- Starting point – heritability of reading skills; dyslexia runs in families

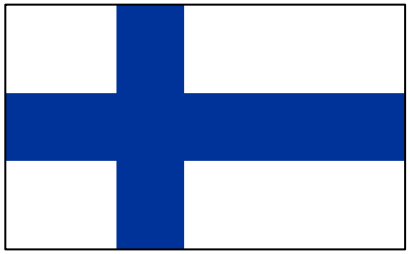
- Recruit children who have a first degree relative with dyslexia
  - Usually a parent (some studies include younger siblings of children with dyslexia)
  - Dyslexia in parent/sib often self-reported

- Examine progress in relation to comparison groups
  - Children from families with no affected relative (TD)
  - Comparison with children with a speech/language impairment

Why Family-Risk Studies?

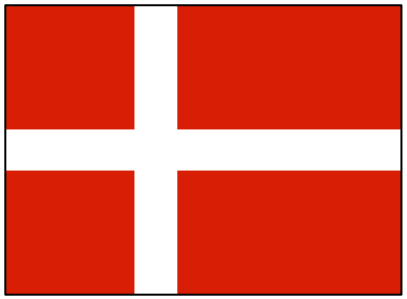
- Follow children from before reading acquisition and therefore are free of sampling bias (relatively!)
- By following children from before reading instruction allow testing of causal hypotheses (especially when coupled with training studies)
- Allow the investigation of how risk factors for disorders combine to impact on literacy development (developmental trajectories)
- Allow an examination of gene x environment interactions
- Clarification of language-universal versus language-specific manifestations of dyslexia

Finnish

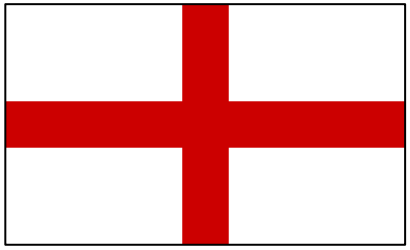


Dutch

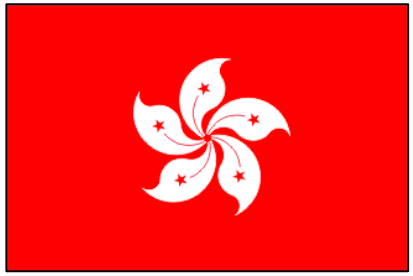
Danish



English



Chinese



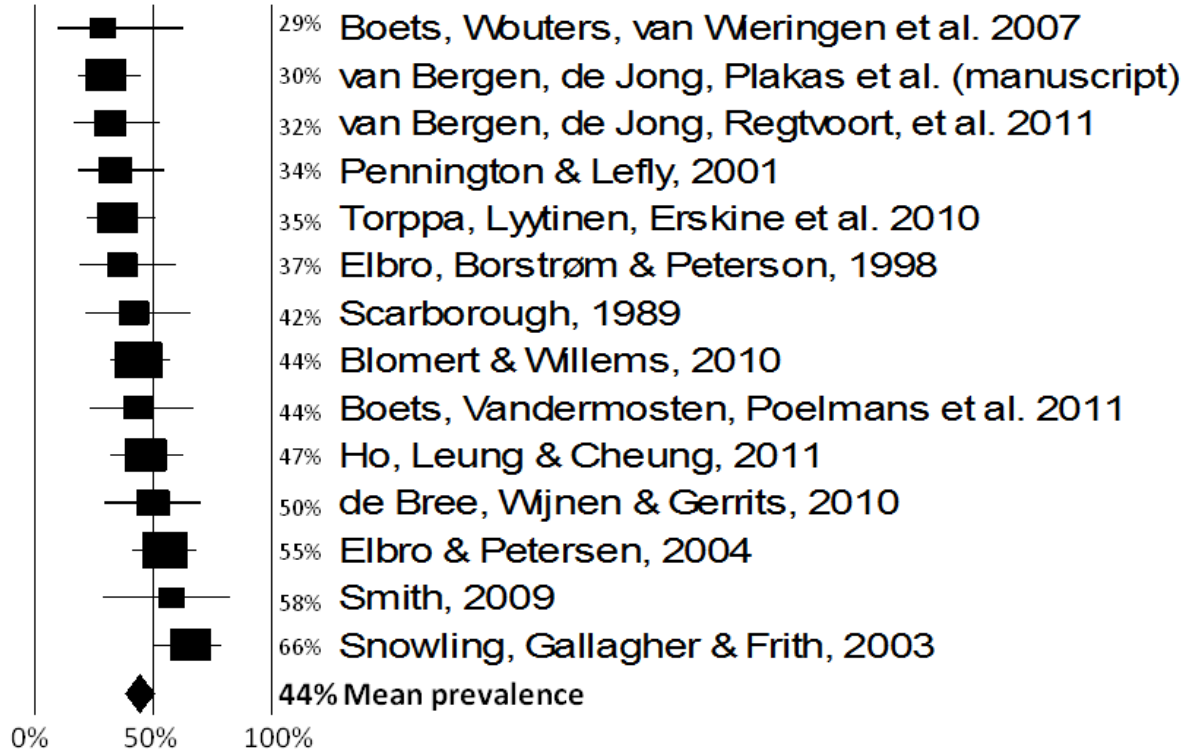
shallow

alphabetic

deep

logographic

Prevalence of Dyslexia

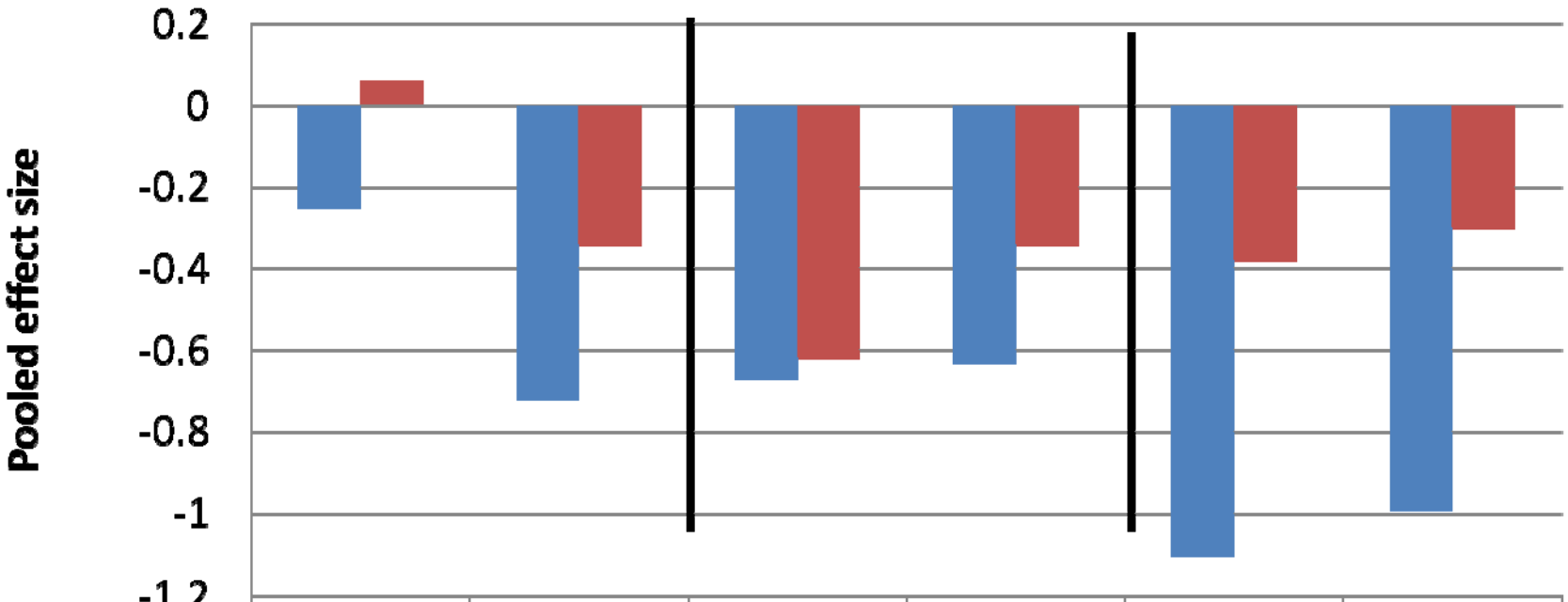


- Cut-off for diagnosis affected prevalence (<10th centile = 35%; <15th = 53%)
- Dyslexia in parents confirmed by test results (45%) cf self-report (40%)

Findings of FR studies

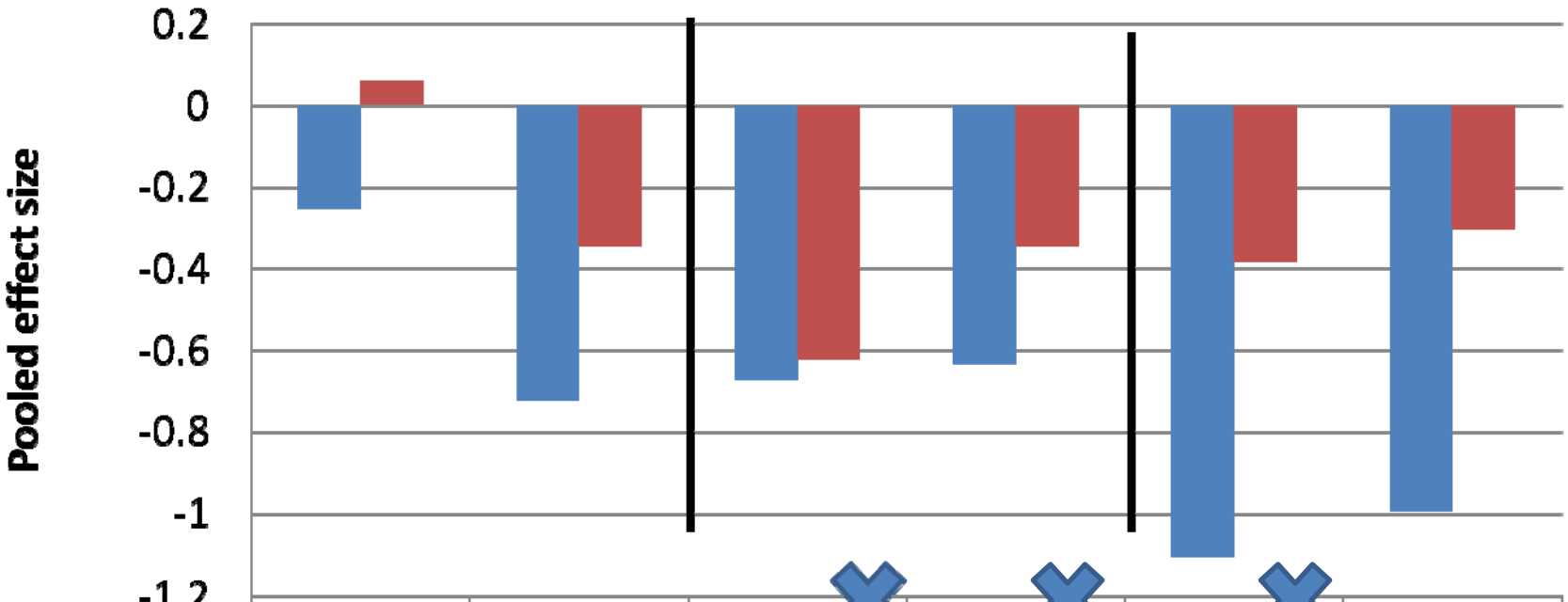
- Classification of participants into groups usually around Grade 2
  - FR-dyslexia
  - FR-Normal Reader
  - TD control (not-at-risk; low-risk)
- Retrospective analysis of group and sub-group differences at earlier developmental stages: precursors
- Meta-analysis of 7 studies

FR: Preschool Skills



	IQ	Lang Comp	PMem	PA	RAN	LK
Dys/TD	-0.25	-0.72	-0.67	-0.63	-1.1	-0.99
FR_NR/TD	0.06	-0.34	-0.62	-0.34	-0.38	-0.3

FR: Preschool Skills



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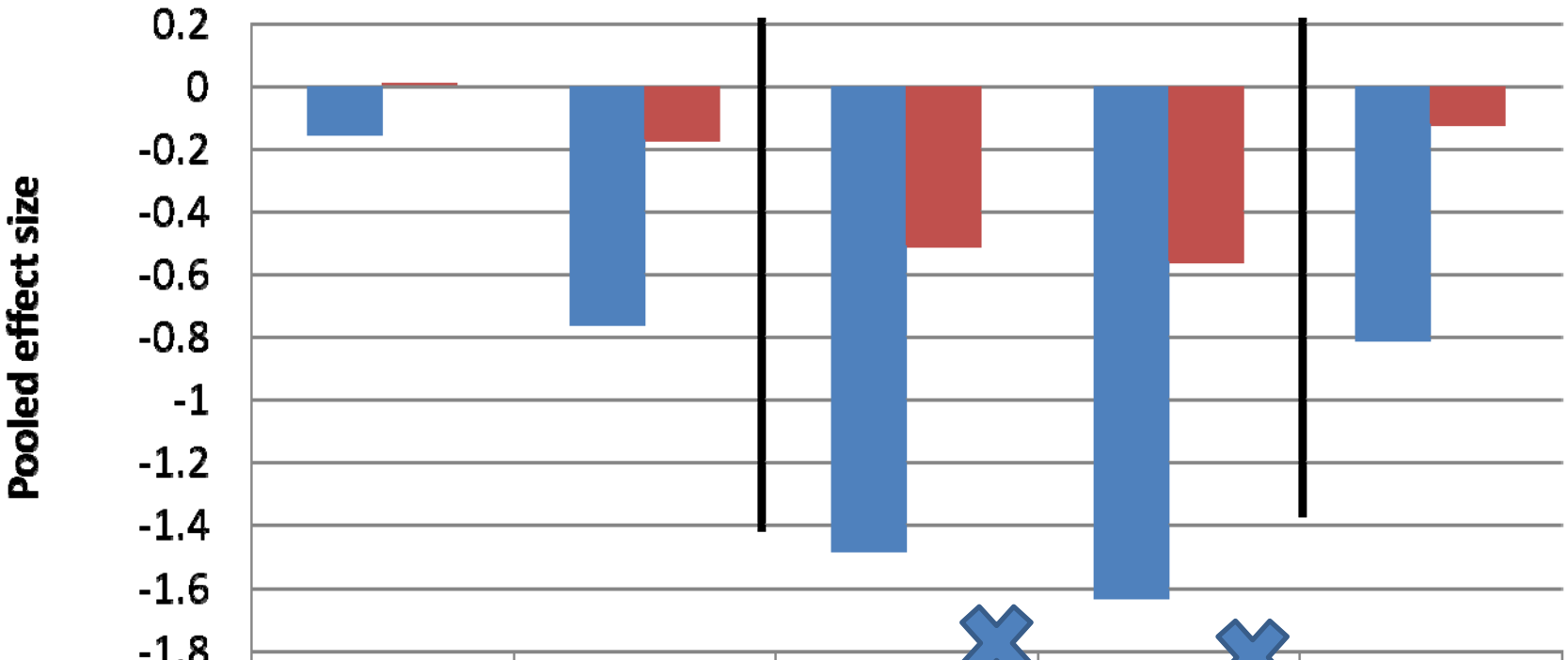
FR: Primary School Skills



	IQ	Lang Comp	PMem	PA	RAN
Dys/TD	-0.15	-0.76	-1.48	-1.63	-0.81
FR_NR/TD	0.01	-0.17	-0.51	-0.56	-0.12

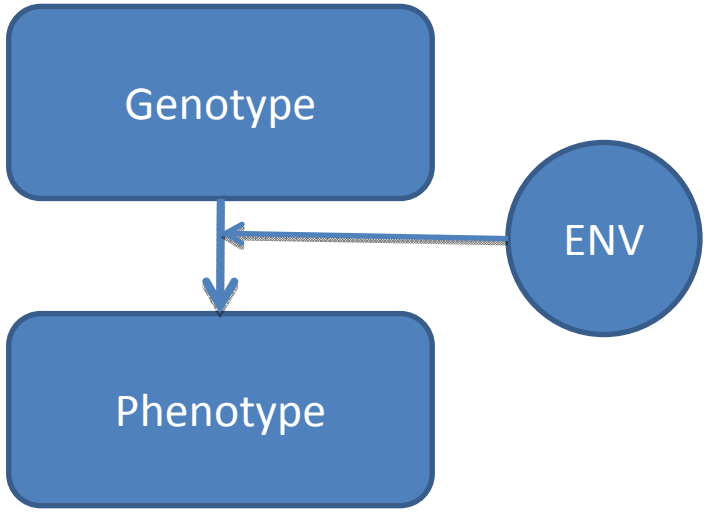
Summary: Meta-Analysis

- Differences in the outcomes of FR and not-at-risk children are not associated with differences in nvIQ
- FR children who become 'dyslexic' show deficits in language comprehension, phonological skills, RAN in pre- and primary school
- FR 'normal readers' show deficits in phonological skills at both ages
  - phonological deficit is a 'marker' of dyslexia risk [endophenotype]
- The phonological deficits of the two FR groups are similar in magnitude during pre-school but greater for the 'dyslexic' group in primary school
  - possible reciprocal influence of letter knowledge/literacy on phonological development (Nation & Hulme, 2010, *Psych Sci*)

Dyslexia: A Causal Chain

- *Genotype* 'The genetic makeup of an organism or an individual'

- *Phenotype* 'The observable characteristics of an individual resulting from the interaction of its genotype with the environment'



Endophenotype 'A marker of a disorder that is intermediate between the genotype and the phenotype'

Dyslexia

- *Genotype* 'The genetic makeup of an organism or an individual'

SUSCEPTIBILITY GENES

- *Phenotype* 'The observable characteristics of an individual resulting from the interaction of its genotype with the environment'

BEHAVIOURAL MANIFESTATION

- *Endophenotype* 'A marker of a disorder that is intermediate between the genotype and the phenotype'

COGNITIVE RISK FACTOR

Endophenotypes

- Endophenotypes have 3 main characteristics
  - Heritable (influenced by susceptibility gene/s)
  - Associated with the deficit in the population
  - State independent: present in unaffected individuals but potentially reduced in severity
    - (Breadon & Freimer, 2006)

- Meta-analysis suggests a phonological deficit is an endophenotype of dyslexia

- The effect of this endophenotype may be moderated by other risk and/or protective factors [language comprehension; RAN]

wellcome trust

Wellcome At-Risk Project

Children at-risk of reading difficulties

-Family Risk

-Pre-school LI

-Children at low-risk of RD

What Risks Do FR Parents Confer?

- 417 parents (238 Mo; 179 Fa) completed a questionnaire probing their literacy skills (adapted from Smythe & Everatt's ADQ)
  - *Do you think you are dyslexic?*
- An adult ADHD screening test (ASRS; Kessler et al., 2005)
- 383 (92%) completed self ratings of language and communication difficulties (CC-SR; Bishop, Whitehouse & Sharp, 2009)
- 344 (82%) agreed to psychometric testing

	TD group	FR group
Mean Literacy (SS)	107.54 (10.6)	93.85 (14.6)
Self Report Dyslexia	n/a	48%
Literacy Skills <90	3.2%	35%
Language Impairment	5.7%	14.8%
High ADHD rating	14.6%	26.4%

Family Risk is not just of 'dyslexia' but extends to related disorders

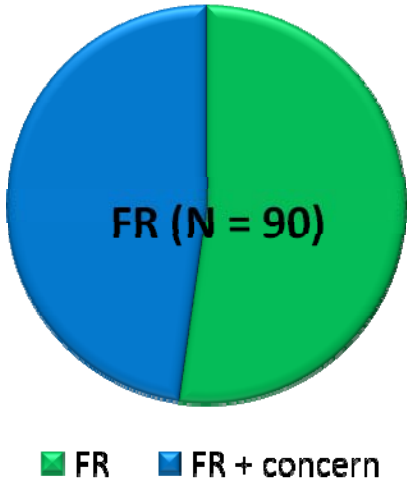
Additional Risks: Language Impairment and Attention Difficulties

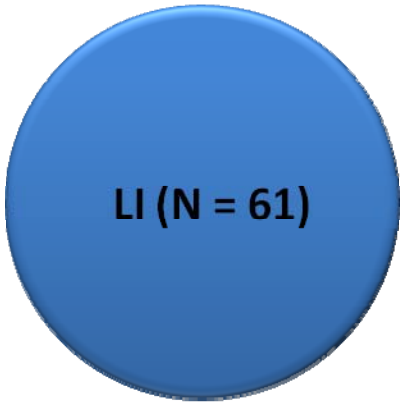
Oct 2007 Recruitment began	**March 2008 Phase 1 began**	**Feb 2009 Phase 2 began**	Feb 2010 Phase 3 began	March 2011 Phase 4 began	Phase 5

3;06 **7;06**

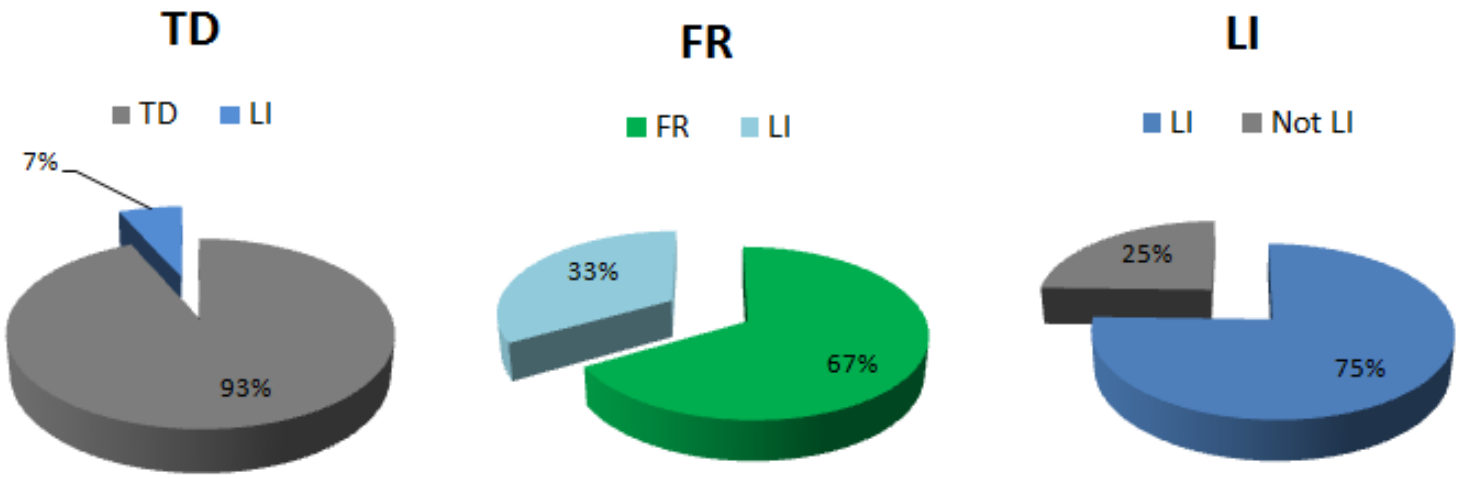
Intervention for 60 poorest readers

WE ARE HERE!



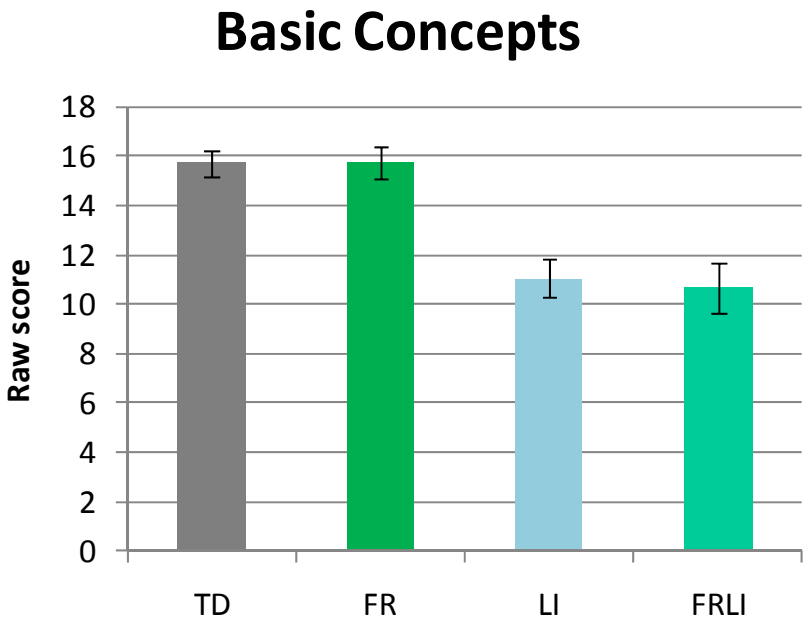


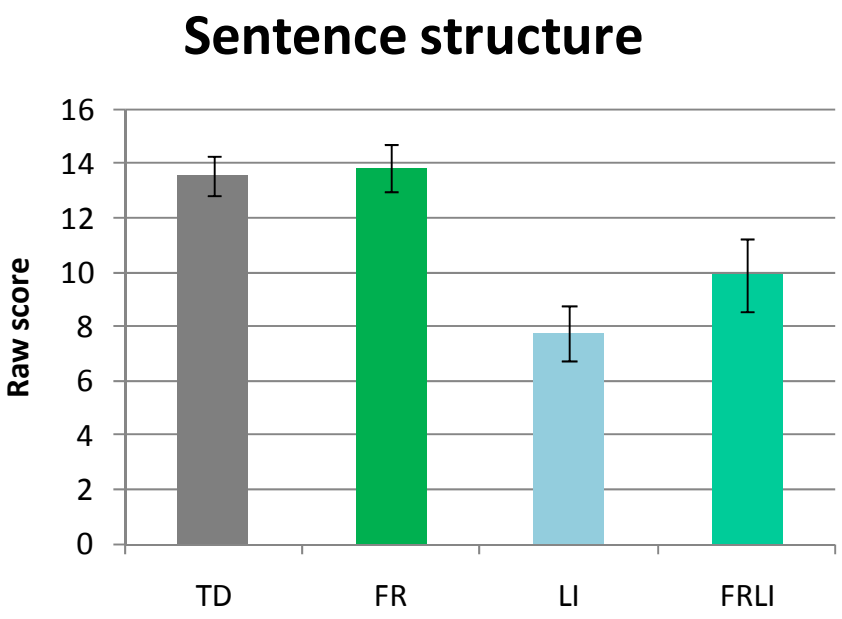
Diagnostic criteria for LI



LI = below SS 85 or criterion on 2/4 language tests
(CELF BC, EV, SS + TEGI)

Receptive Language





CELF Basic Concepts

Point to the one that's long (child sees three pencils – one long, one short and one in between

(TD = FR) > (FRLI = LI)

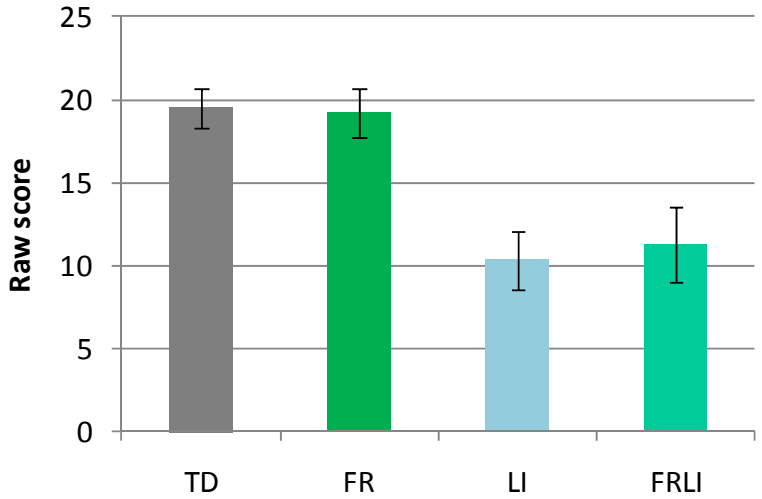
CELF Sentence structure

Point to 'the girl is being pushed by the boy'

(TD = FR) > FRLI > LI

Expressive language

Expressive vocabulary

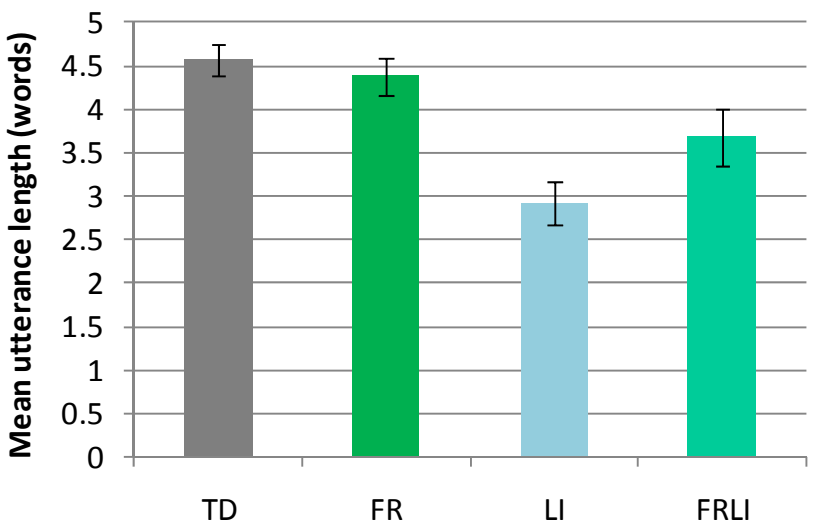


CELF Expressive Vocabulary

Picture naming

(TD = FR) > (FRLI = LI)

MLU

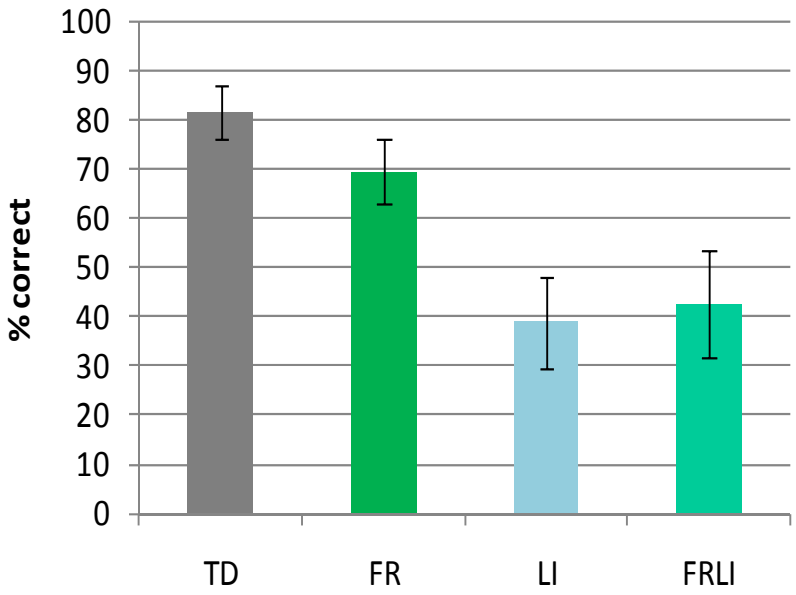


Natural Language Sample

(TD = FR) > FRLI > LI

Morpho-syntax

Past tense

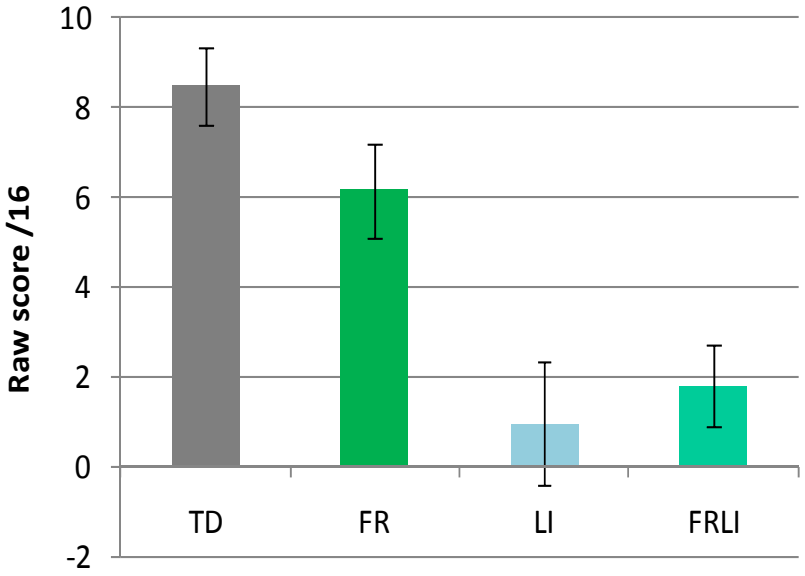


TEGI (verb inflection)

Here the boy is painting (picture)
now he's done (picture)
tell me what he did…

TD > FR > (FRLI = LI)

Sentence repetition /16

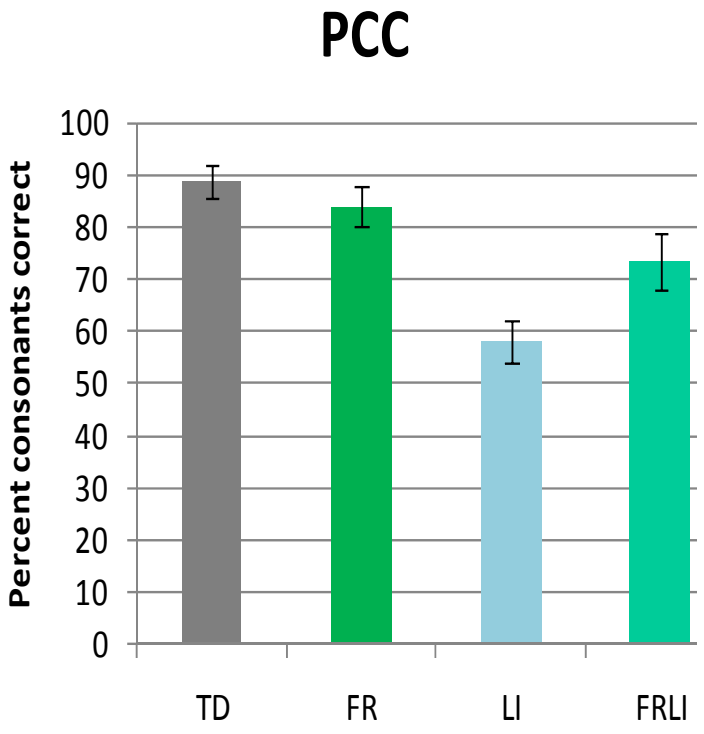


SIT-16

Repeat sentences verbatim

TD > FR > (FRLI = LI)

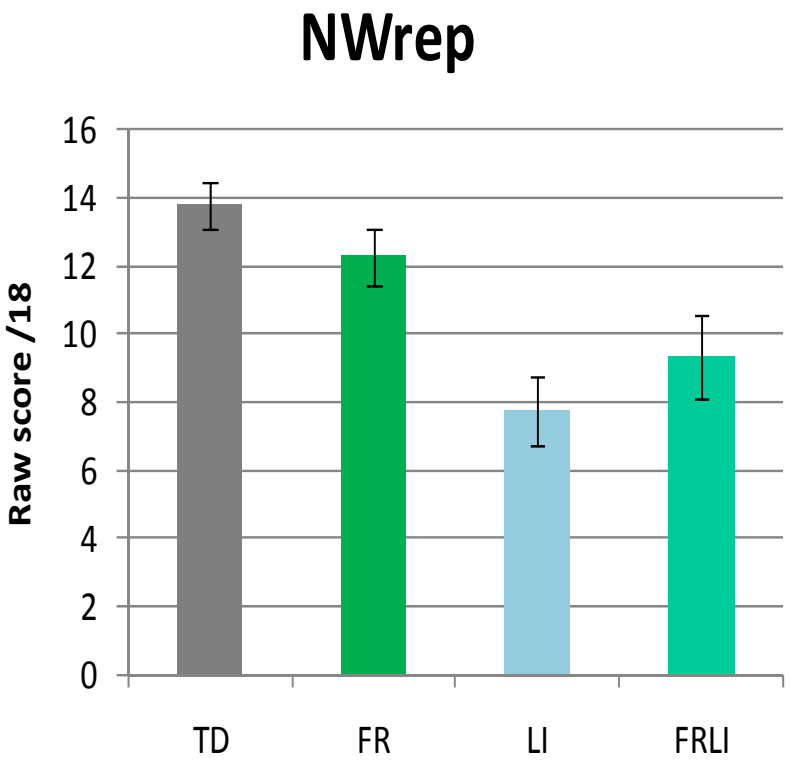
Phonology



DEAP

Articulation test (PCC)

TD > FR > FRLI > LI



PSRep

Repetition

1-3 syllable words and nonwords

TD > FR > (FRLI = LI)

Language Profiles in Preschool Children at FR of Dyslexia

Comparable to TD controls

- Receptive language
- Expressive language skills

Impaired

- Verb inflection
- Sentence Repetition
- Nonword Repetition

A further one third of the FR group fulfil criteria for language impairment

Putative Endophenotypes of Dyslexia

- Children at FR of dyslexia without a clinically significant language impairment showed deficits in:
  - Nonword repetition
  - Verb inflection
  - Sentence repetition

PHONOLOGICAL ENDOPHENOTYPE

- One third with a significant language impairment (FR+LI)

LANGUAGE (comprehension) ENDOPHENOTYPE

Siblings in the FR group

Centre for Reading and Language

THE UNIVERSITY of York

Participants

- 84 siblings
  - 63 FR (high-risk)
  - 21 TD (low risk)
- Crtierion: WIAT reading or spelling SSc ≤ 85 or a diagnosis of dyslexia

	FR – Dys N=32	FR – NR N=31	Controls
Age [months]	114 (24)	98 (20)	95 (18)
Full IQ	104 (15)	114 (16)	119 (14)
WIAT reading	83 (13)	107 (10)	108 (9)
WIAT spelling	81 (10)	106 (13)	105 (11)

Language Tasks

Derivational Morphology

She plays music, so we can say she is a _____ (musician)

He is very strong, so we can say he has a lot of_____ (str*ength*)

PA (after McDougall, Hulme, Ellis, & Monk,1994)

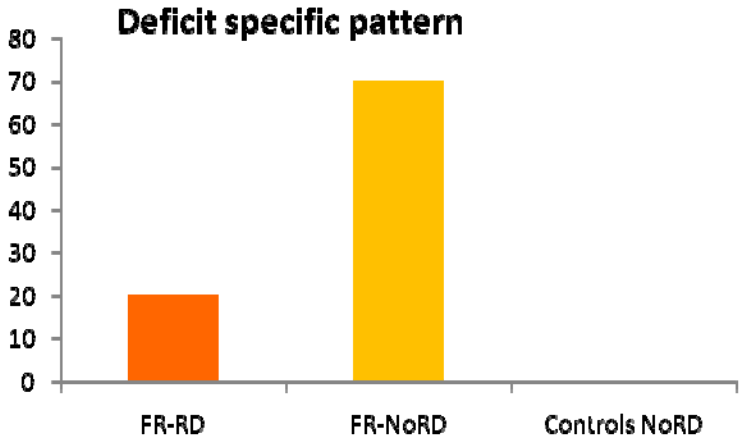
Phoneme deletion task

Phonological Memory (adapted version of nonword repetition task; Dollaghan & Campbell,1998)

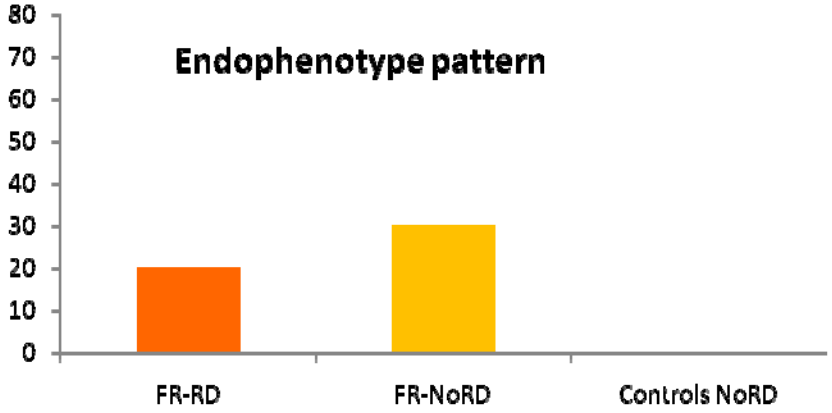
RAN (van der Sluis et al. 2004)

Digits

Rationale

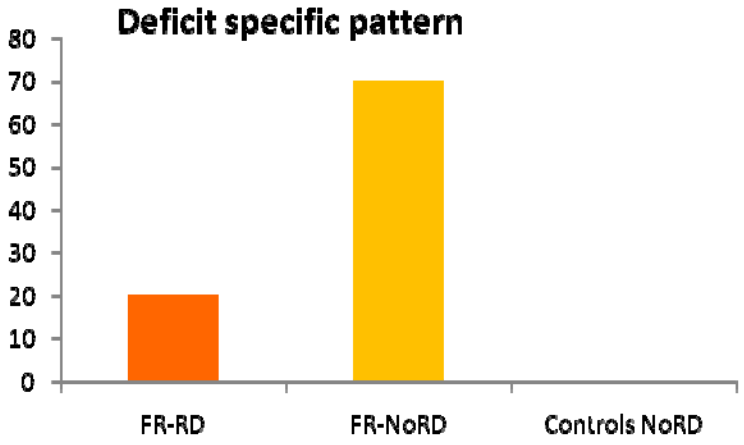


- Only dyslexic group show a deficit

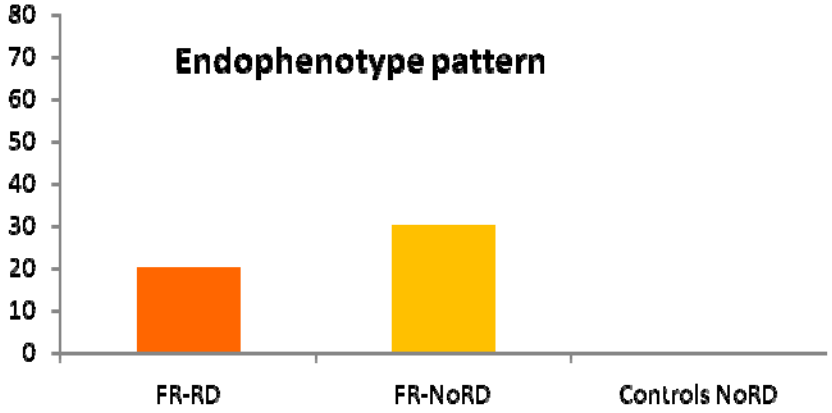


- Both FR groups show deficit, varying in severity

Findings



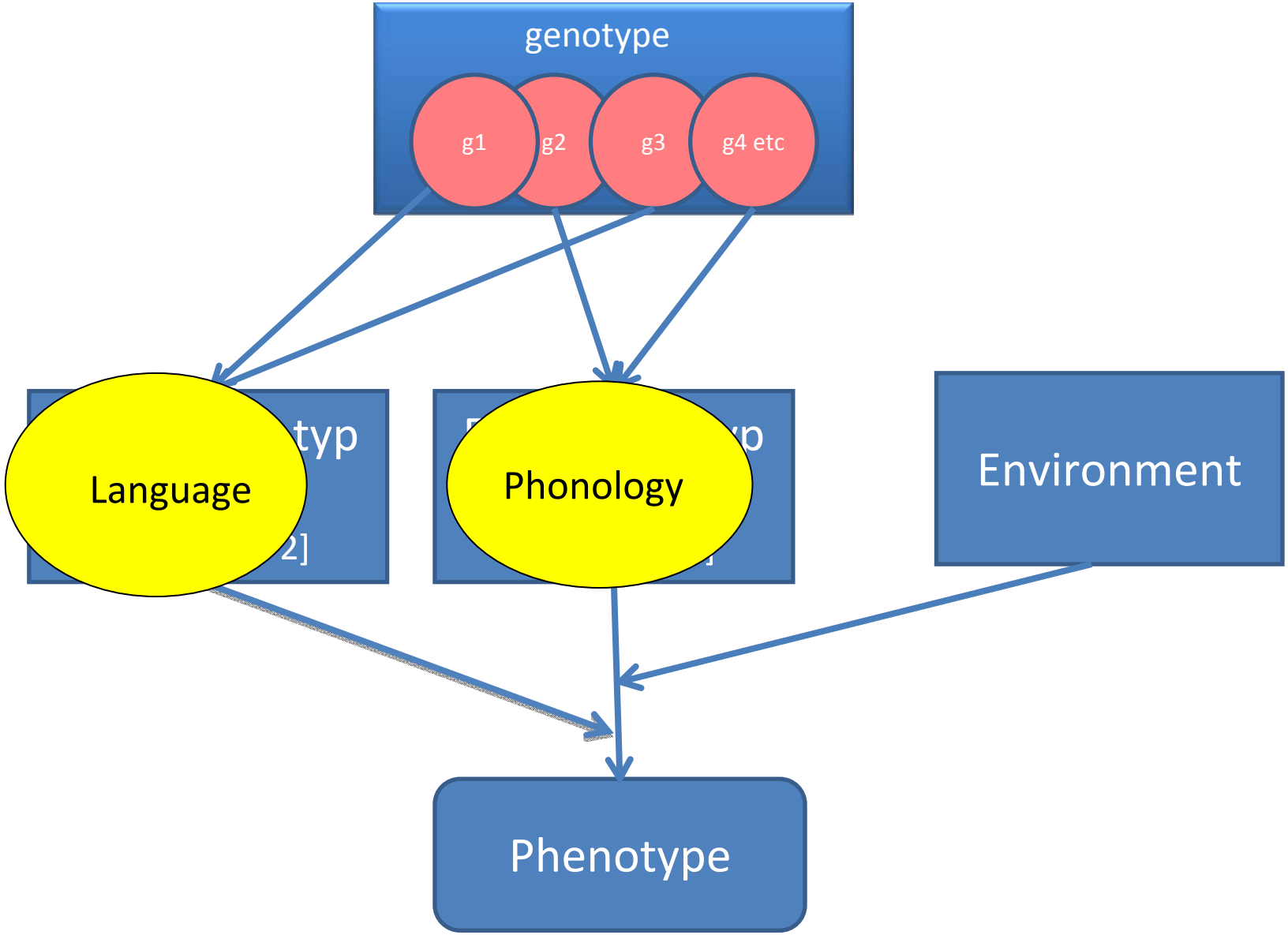
- Language (Morphology)
- RAN

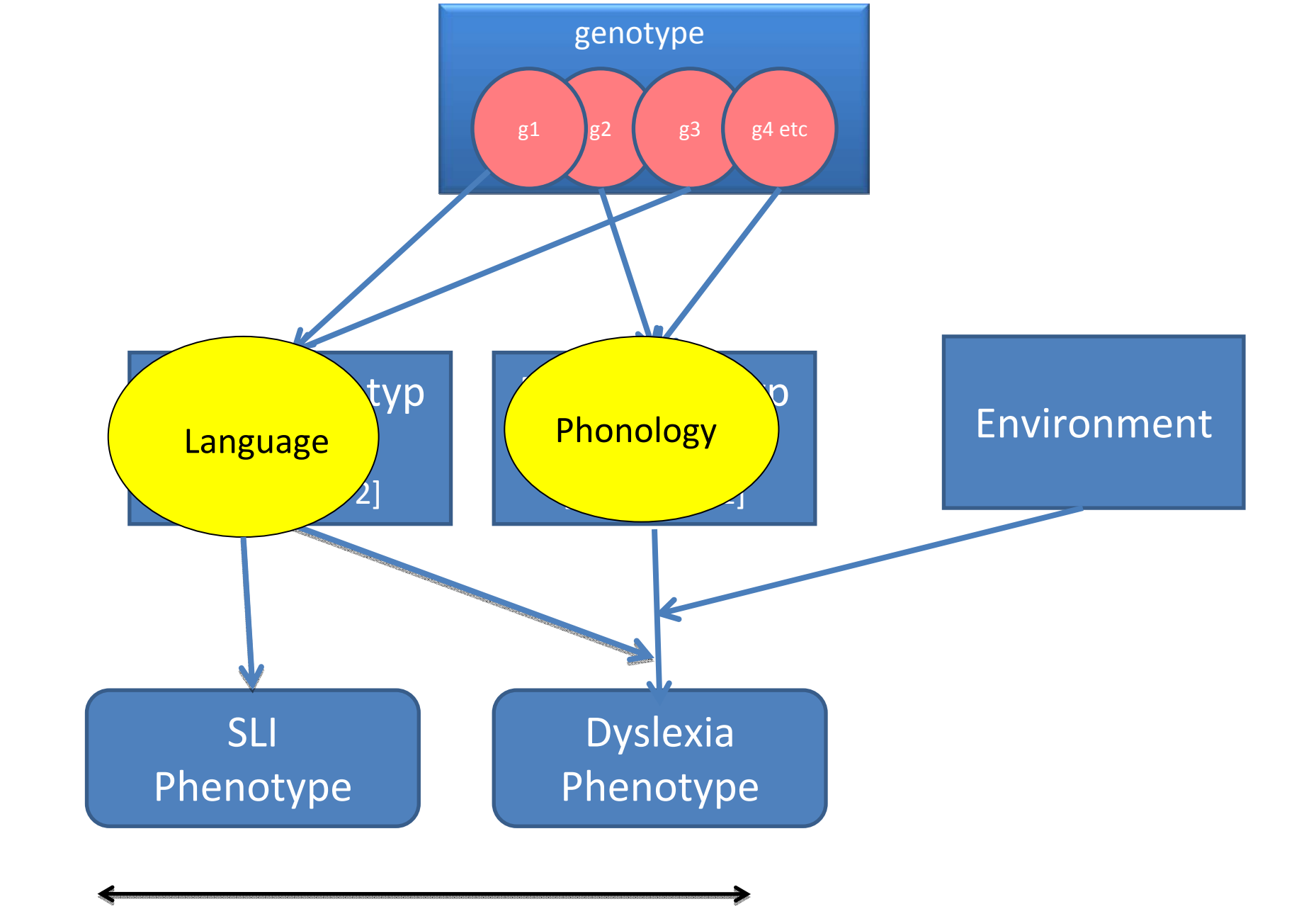


- Phonological Memory
- PA ++

Summary: FR Study

- At pre-school, two putative endophenotypes:
  - Phonological Deficit (PM and PA)
  - Language Deficit (comprehension?)
- At school age, dyslexia associated with both Phonological and Language endophenotypes
- Parents confer risks for both poor literacy (?P = risk 1) and poor language (L = risk 2)
  - Poor P -> dyslexia
  - Poor L -> SLI


genotype
g1
g2
g3
g4 etc
Language
Phonology
Environment
Phenotype


genotype
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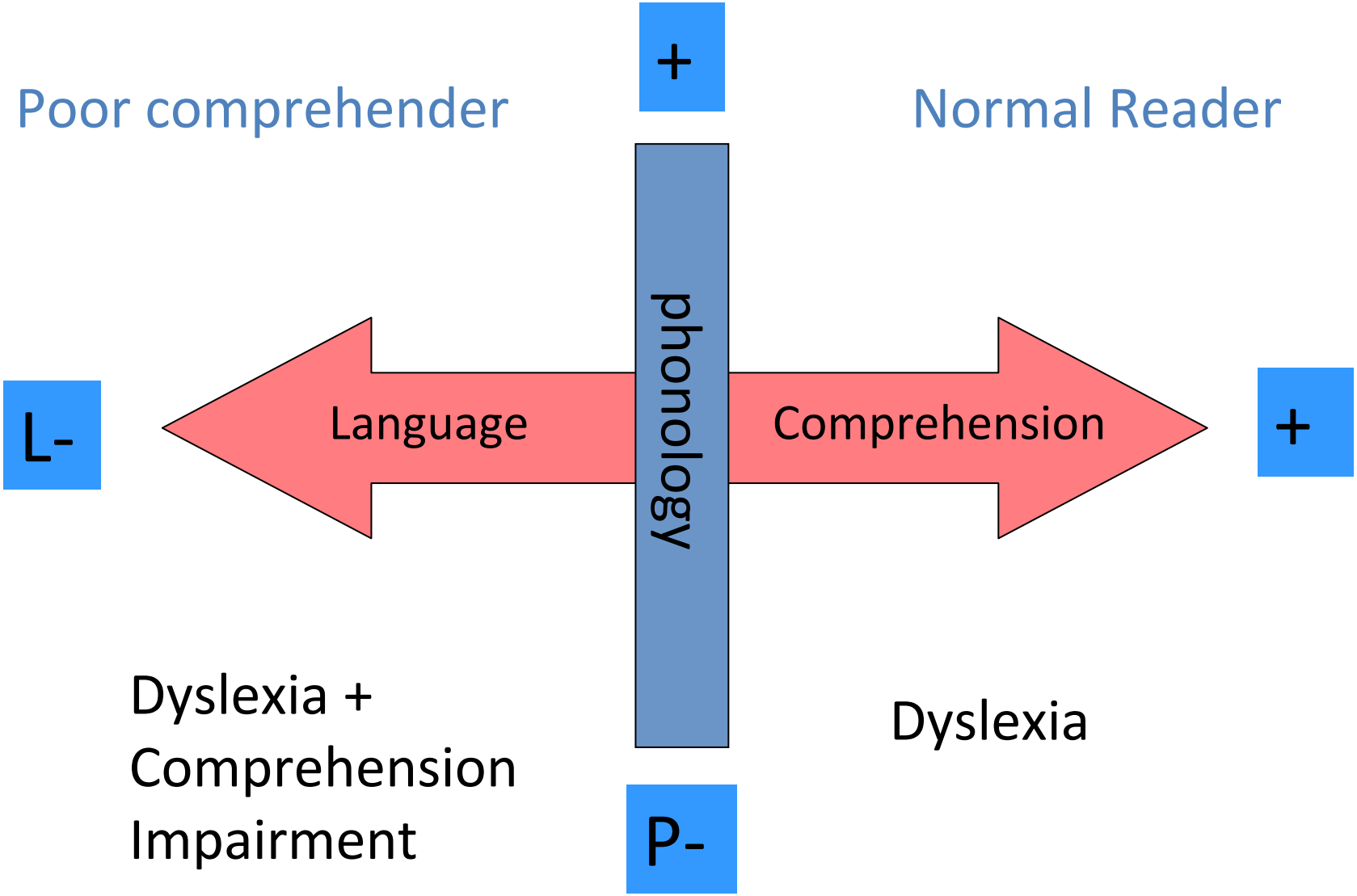
Dyslexia
Phenotype

Spectrum of Reading Disorders

(after Bishop & Snowling, 2004)



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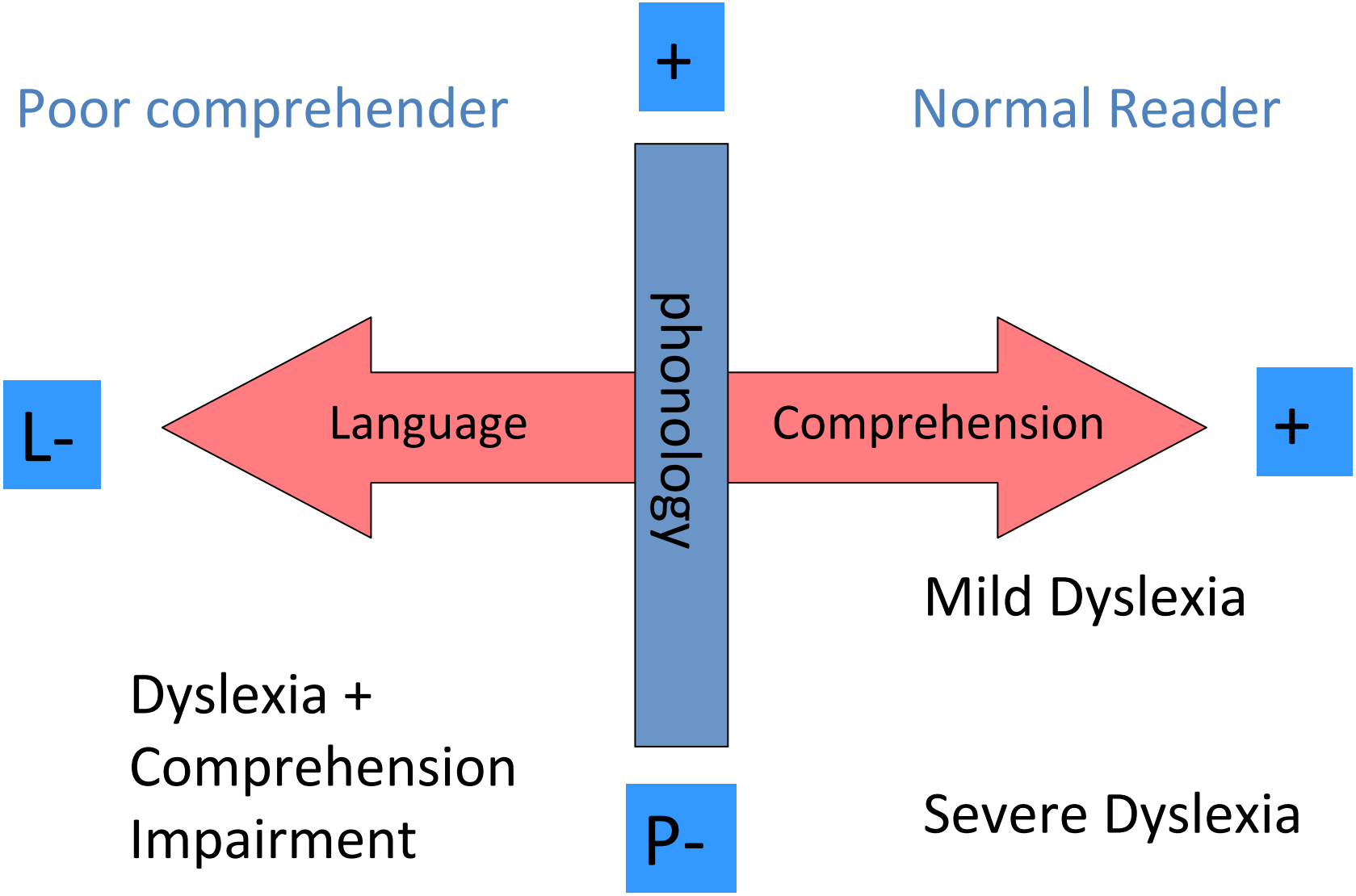


‘It’s the *environment* stupid!’ (Sonuga-Barke, 2010)

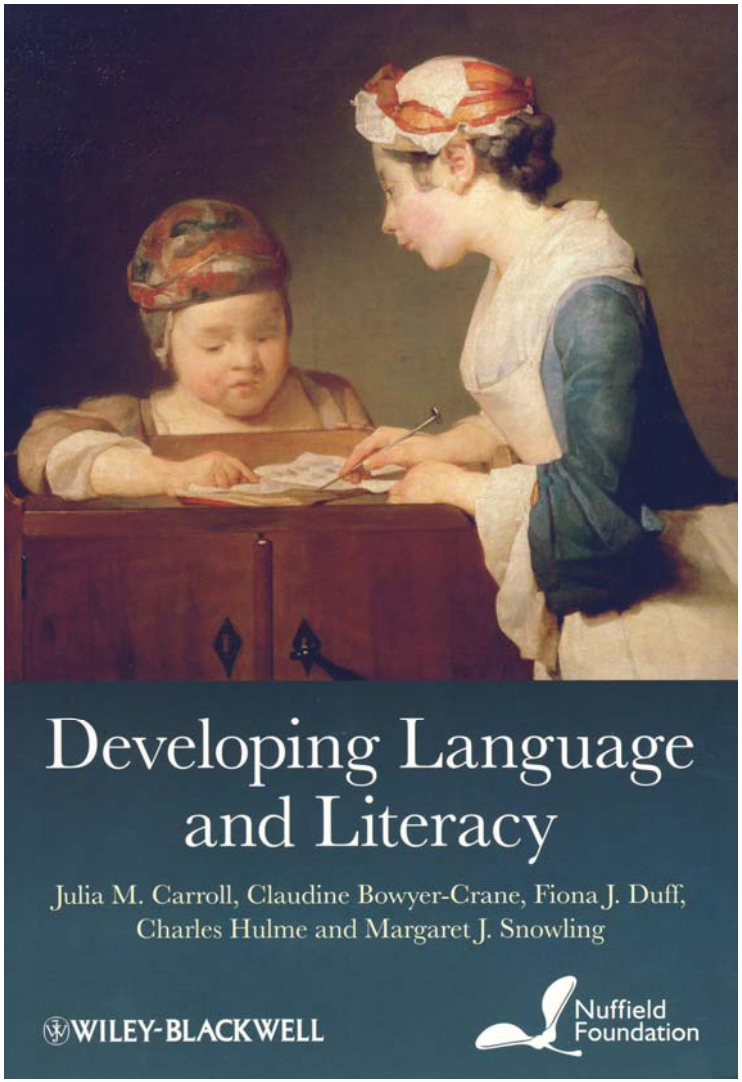
- **‘we can at last bypass this ...nature vs. nurture ‘dogma’. Serious science is now more than ever focused on the power of the environment to shape neuro-developmental processes and pathways’**
  - Home Literacy Environment
  - Intervention
  - Language of instruction

Home Literacy Environment (HLE)?

- In our study, the families of preschool FR and TD children provide similar literacy environments for their children
  - Shared Reading
  - Direct Literacy Instruction
- In the prediction of pre-school letter-knowledge
  - direct instruction accounts for 19% of the variance for the FR group
  - only 3% for the low risk TD group

Implication: parental input pays off at least with regard to foundation literacy skills

Intervention?



- Children with poor phonological skills benefit from intervention that includes training in phoneme awareness linked with reading instruction
- Children with language difficulties benefit from oral language interventions which in turn foster reading comprehension
- Such interventions can be delivered effectively in Early Years settings – Charles Hulme's talk

Language of Instruction?

Czech Kannada Slovak

Conclusions

- Language (comprehension) and phonological skills are two dimensions underpinning literacy development

- FR Studies
  - phonological deficits – an 'endophenotype' of dyslexia which confers risk of reading disorder
  - language skills –an additional risk factor (endophenotype of SLI?)

- Dyslexia is a dimension, not a clear cut category

- The behavioural manifestation of 'dyslexia' depends upon a complex interaction of risk factors which act through the home and school environment

Implications for Practice

- Since we can do nothing about our genes, then it is ever more important to use our scientific understanding to implement effective interventions that ameliorate literacy difficulties

Thank You!

- To all children, parents, TAs, teachers, schools and SLTs who support our research

- CRL: Wellcome team, ELDEL, Interventions team, Clinical team, graduate students and research assistants

- Funders: Wellcome Trust; Nuffield Foundation; Marie Curie FP7; ESRC; Royal Society, British Academy, GL