What causes language disorders in children?

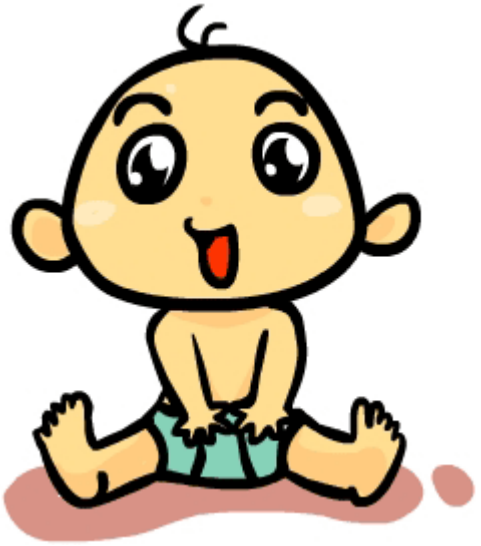
Dorothy Bishop
University of Oxford

Specific language impairment (SLI)

- Diagnosed in children when language does not follow normal developmental course
- Problems with language structure (phonology and syntax) common
- Not due to hearing loss, physical abnormality, acquired brain damage
- Normal development in other areas

What causes SLI?

Theory 1: inadequate input



http://news.bbc.co.uk/1/hi/education/2638889.stm

Thursday, 9 January, 2003, 11:15 GMT

Daily grunt parents hold children back

Not enough talking going on

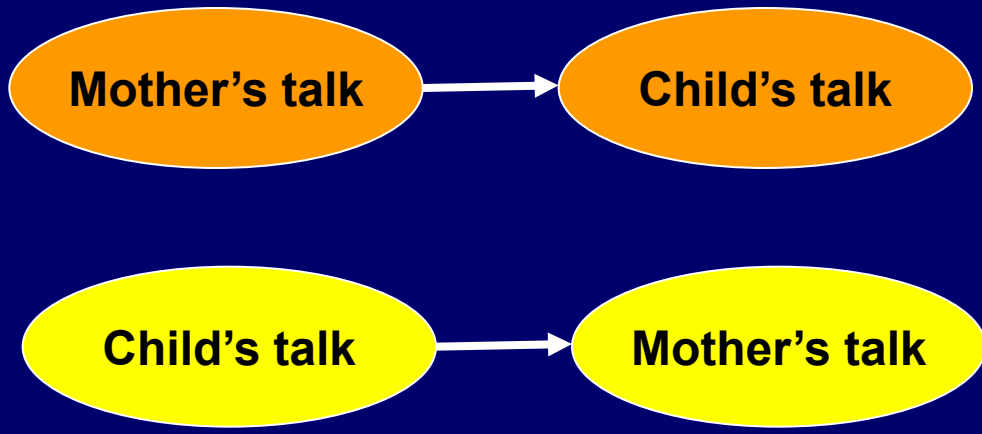
Parents who do little more than grunt at their children every day are damaging their language development, a literacy expert has said.

Alan Wells, director of the Basic Skills Agency, says parents no longer talk to their children and instead just let them sit in front of the television or computer for hours.

How plausible is poor language environment as cause of SLI?

- All agree that to learn a language must hear that language – Danish children learn Danish, English children learn English!
- “Motherese” seen in many (?all) cultures – special way of talking to infants
- Correlations between maternal language and children’s language development

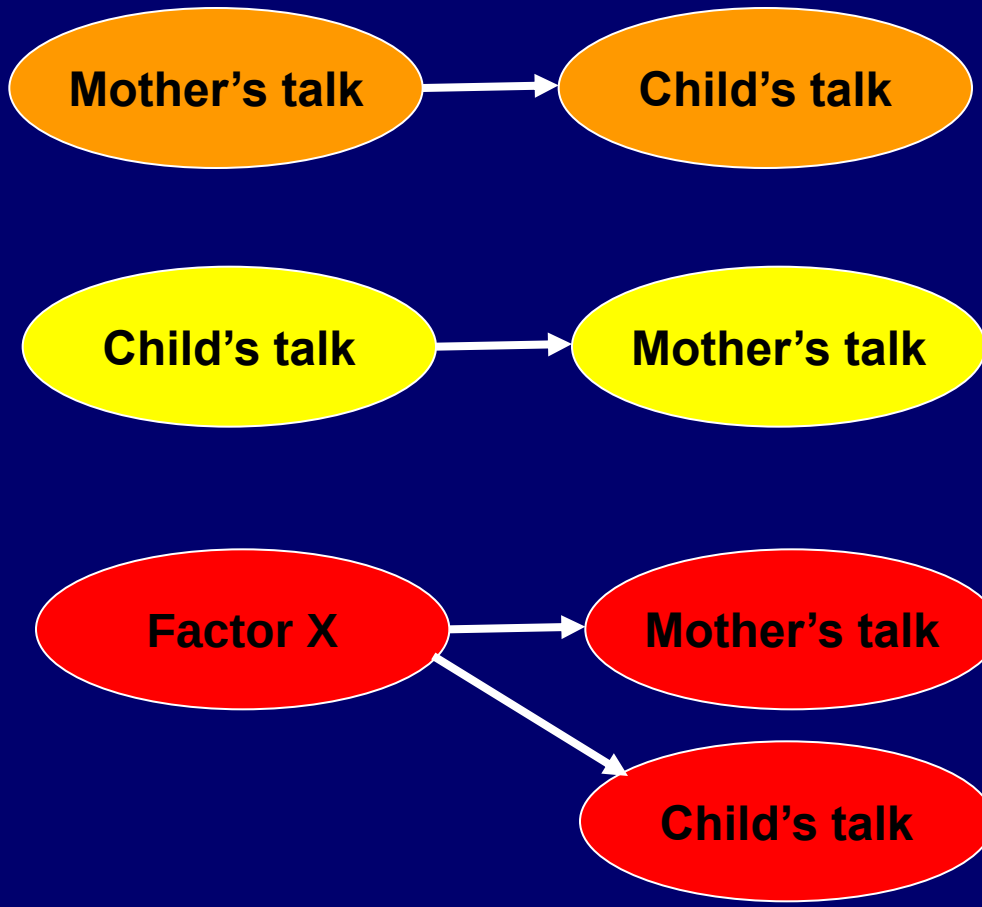
Correlation ≠ Causation



Evidence for evocative effect

- Huttenlocher et al (2007)
  - Study of 50 families, diverse SES
  - Measured quantity, complexity, diversity of parental language to child
  - Quantity did not change with child's age
  - Complexity and diversity increased with child's age

Correlation ≠ Causation

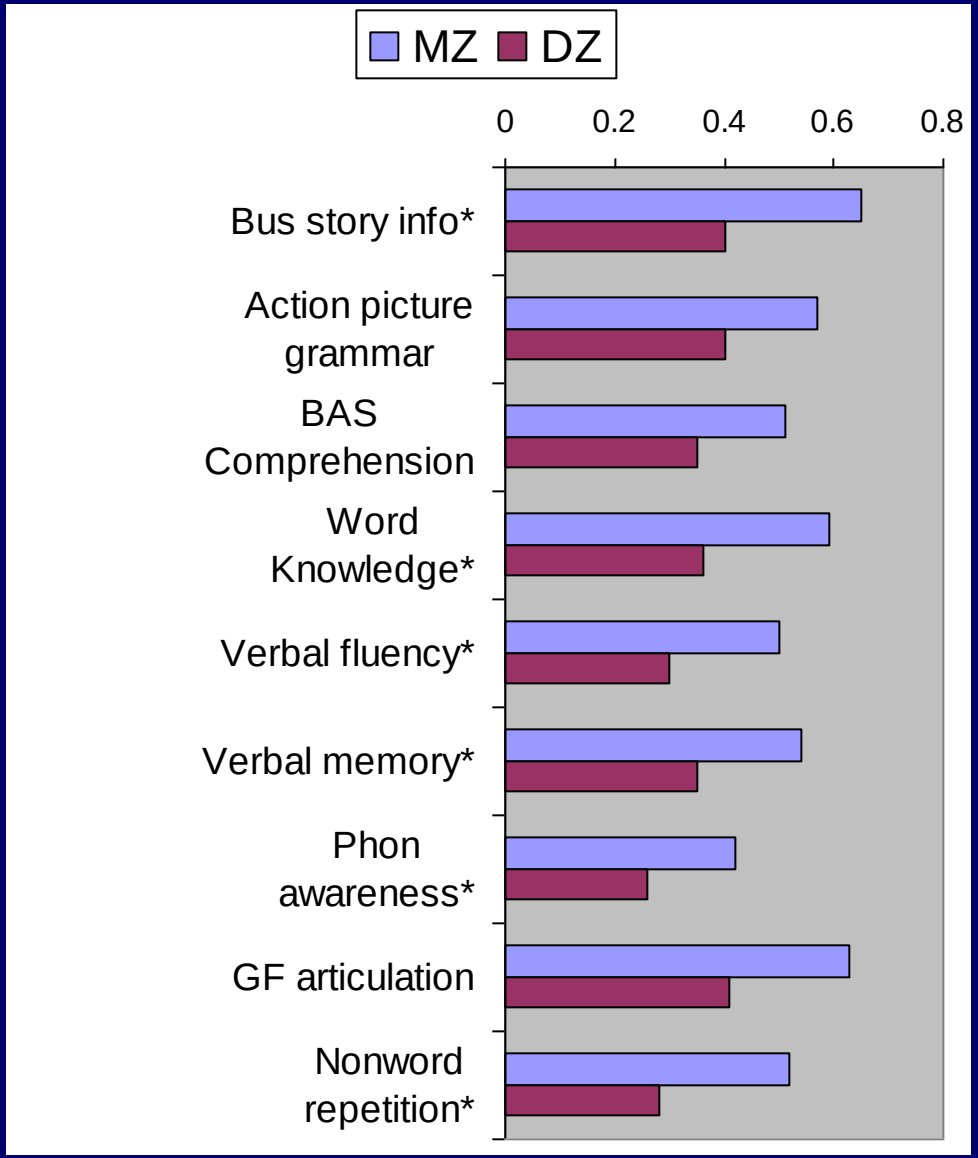


Evidence for non-causal association

- Twins growing up together
  - MZ, genetically identical
  - DZ, non-identical
- If parental language input was important, would expect twins to be similar, regardless of whether MZ or DZ
- For many measures, DZ less similar than MZ

Twin-twin correlations

* Indicates no evidence of environmental influence

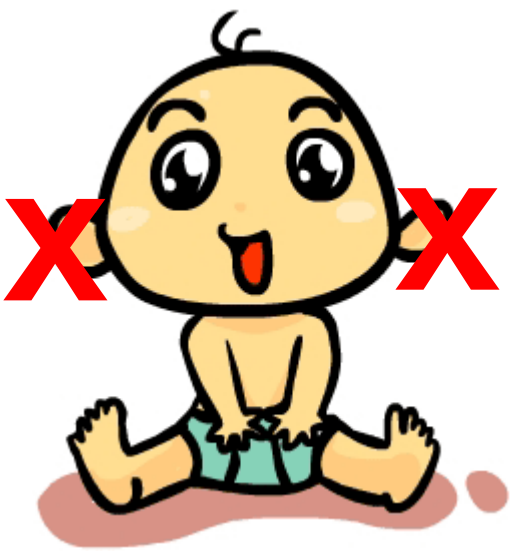


Kovas Y, et al. (2005) Genetic influences in different aspects of language development: The etiology of language skills in 4.5 year-old twins. Child Dev 73: 632-651.

Hearing children of deaf parents

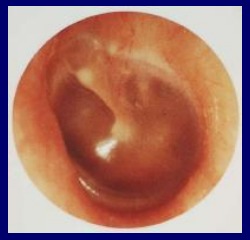
- Schiff-Myers (1988)
  - Either hear no oral language from parents, or hear limited syntax, abnormal articulation/prosody
  - Some learn sign as first language
  - Variable outcomes, but many have no problems with speech and language
  - About 5-10 hr per week exposure to normal speakers seems sufficient

Theory 2: inadequate speech perception



Otitis media with effusion

- **Middle ear fills with fluid**
- **Very common in young children**
- **Causes fluctuating conductive hearing loss**



Healthy tympanic membrane

Chronic otitis media

Otitis media treatment - tympanic membrane hole or perforation with a ventilator tube in place

Otitis media with effusion (OME)

- **Early studies found more language problems in children with OME**
- **But problem of *ascertainment bias***

  - Parent more likely to go to doctor if child has language problems
  - Doctor more likely to recommend treatment if child has language problems
  - Children treated for OME not a typical sample
  - Epidemiological studies: look at whole population – less evidence of language problems

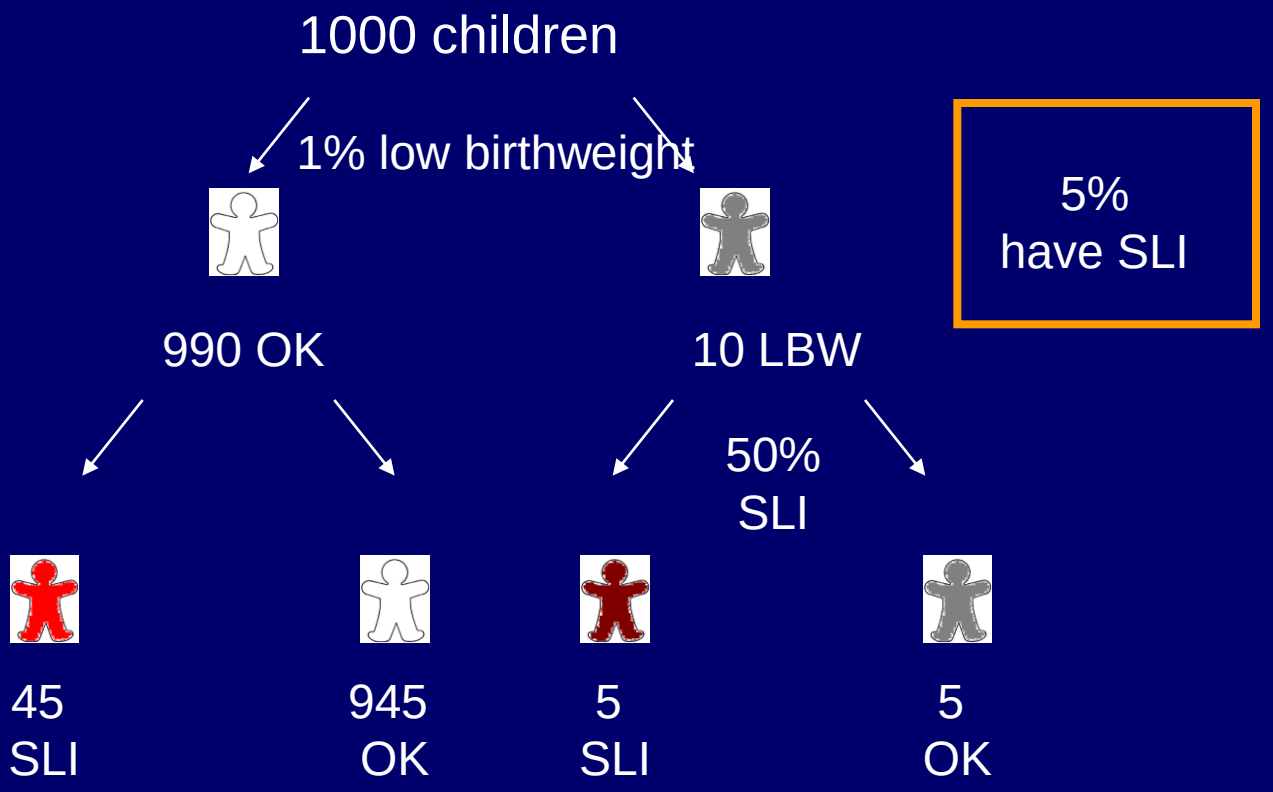
Theory 3: early brain damage

Is brain damage implicated?

- Early idea of “continuum of reproductive casualty” - i.e. brain damage incurred around time of birth might lead to learning disabilities

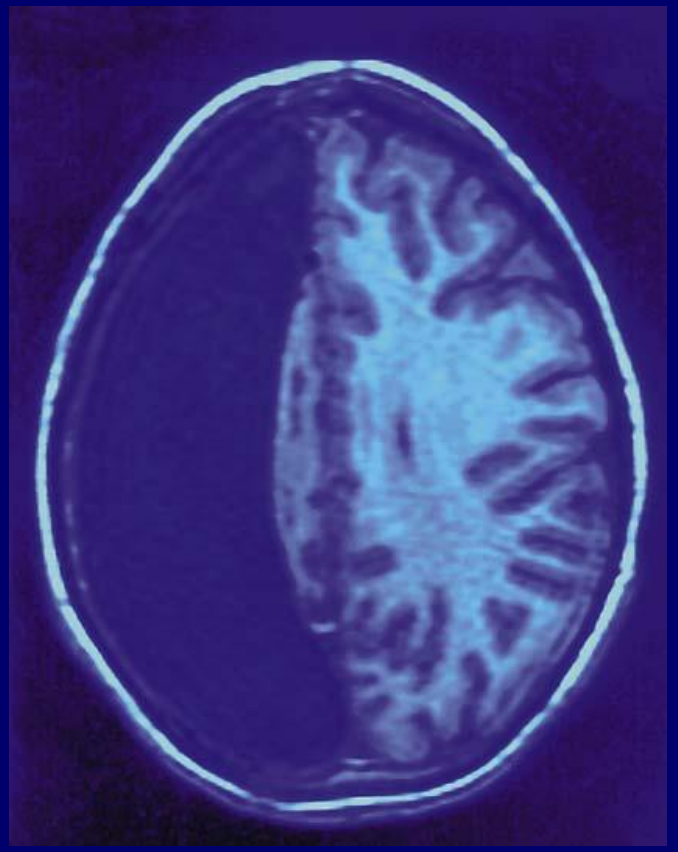
Evidence against brain damage as cause of SLI

- Children with very low birthweight do have increase in all neurodevelopmental disorders, including language
- But, no obvious signs of neurological impairment in most cases of SLI
- And no excess of perinatal problems in SLI


1000 children
1% low birthweight
990 OK
10 LBW
50%
SLI
45
SLI
945
OK
5
SLI
5
OK
5%
have SLI

Evidence against brain damage as cause of SLI

- Children who *do* have focal lesions affecting the language areas don't develop SLI - see Basser 1962!



Theory 4: abnormal neurodevelopment

Clark & Plante, 1998

- 20 biological parents of language impaired children
  - 15 had evidence of residual language difficulties
- 21 unrelated adult controls
  - 4 had evidence of residual language difficulties
- MRI scan of inferior frontal gyrus

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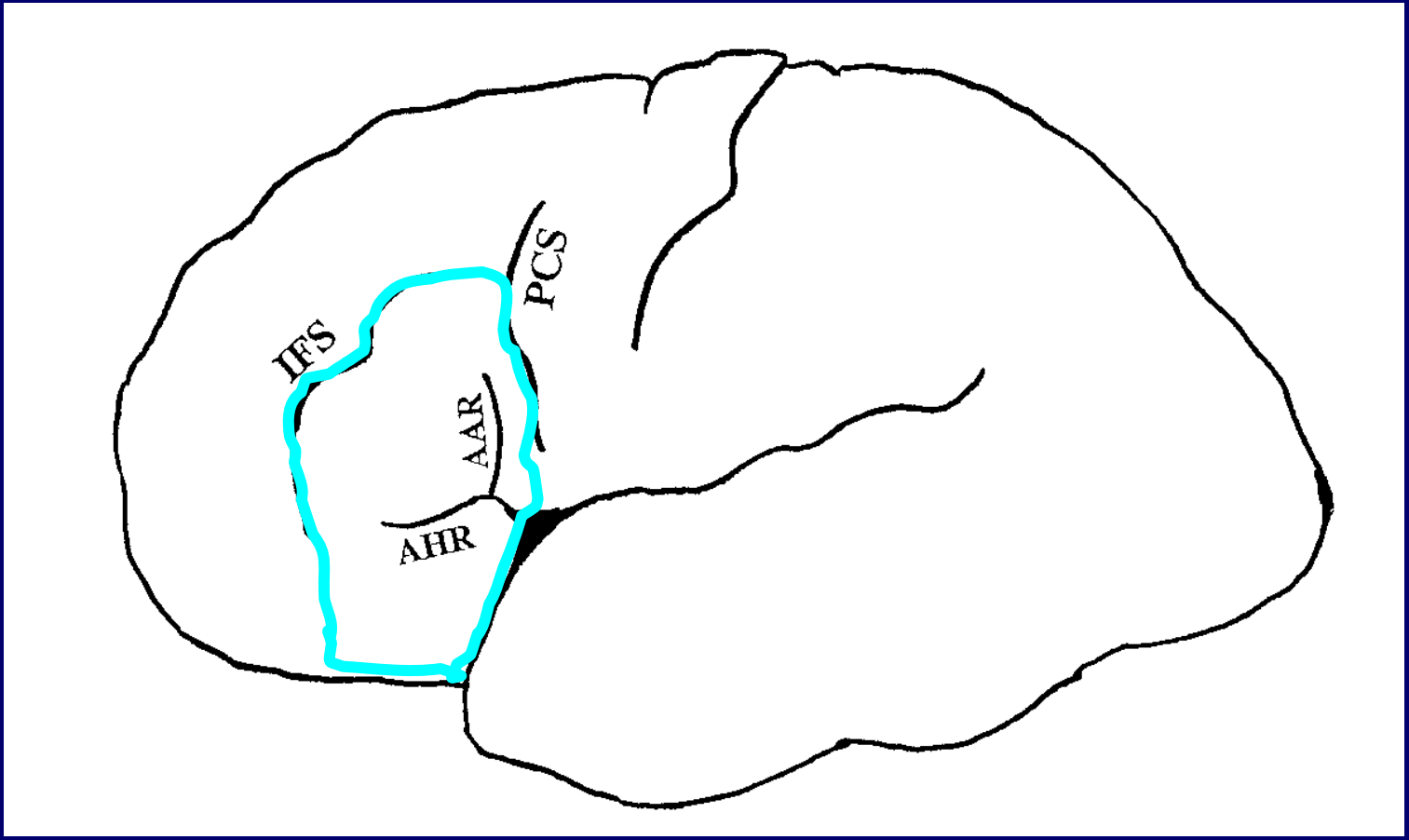

IFS: inferior frontal sulcus
AAR: anterior ascending ramus
AHR: anterior horizontal ramus
PCS: precentral sulcus

from Clark and Plante. 1998 Brain and Language 61, 288-303

Clark & Plante, 1998

N with extra sulcus (either side):

- **With +ve family history**
  - Parent has typical language: 5/10 = 50%
  - Parent has language problems: 20/30 = 67%
- **With no family history**
  - Parent has typical language: 13/34 = 38%
  - Parent has language problems: 6/8 = 75%

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Clark & Plante, 1998

Conclusions

- **Suggests prenatal event has systemic effect on developing brain**

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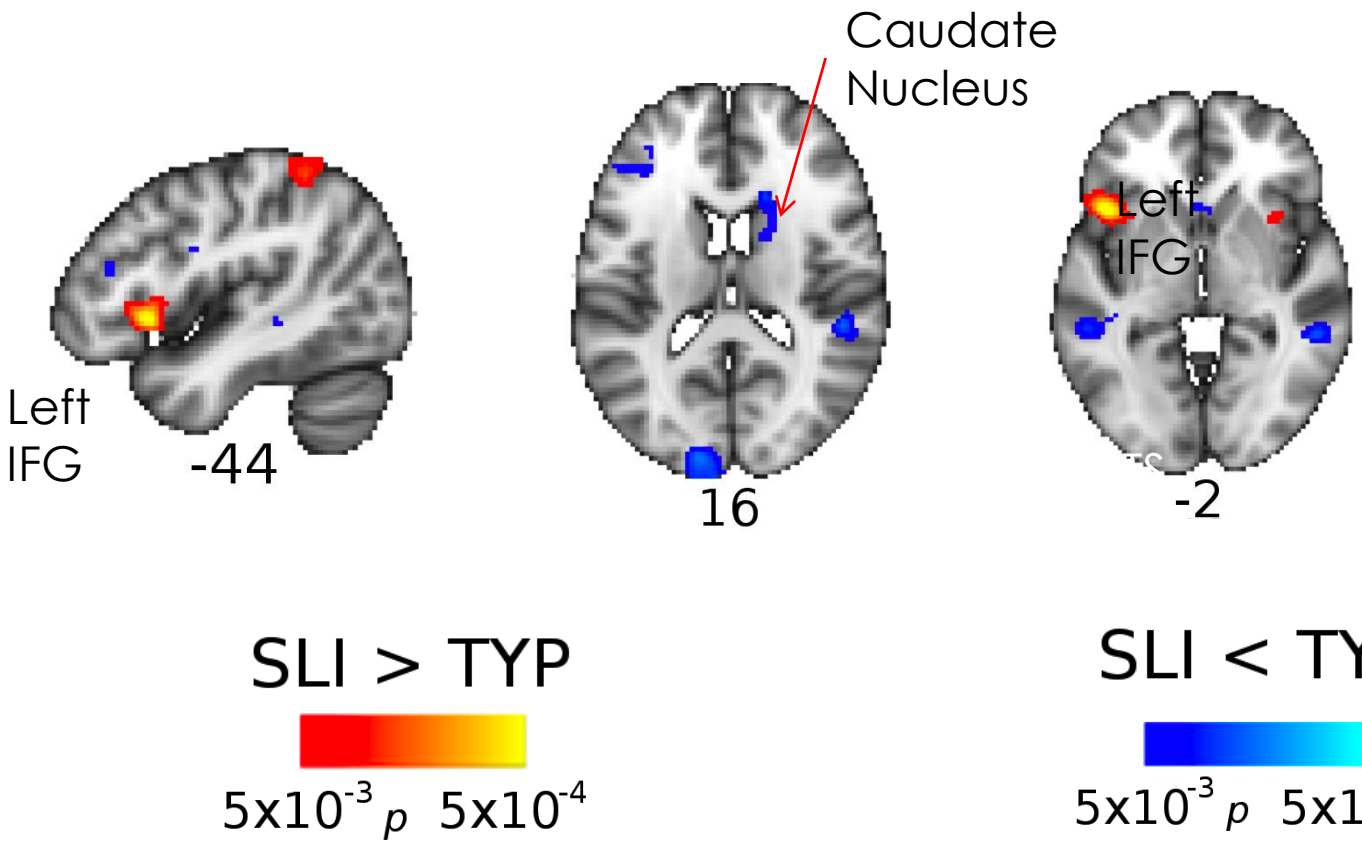
Study of SLI using brain scanning

Kate Watkins

- Oxford study of children with SLI and their families
- No gross differences seen in the brain
- Subtle differences in language areas in distribution of grey matter
- Less activation of language areas when doing a language task

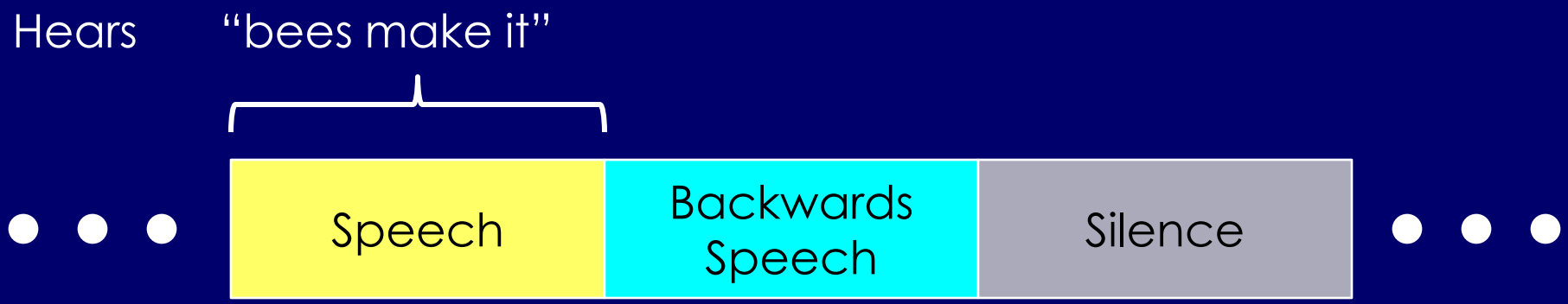
VBM Grey Matter
SLI vs. Typical



More grey matter in left inferior frontal gyrus in SLI

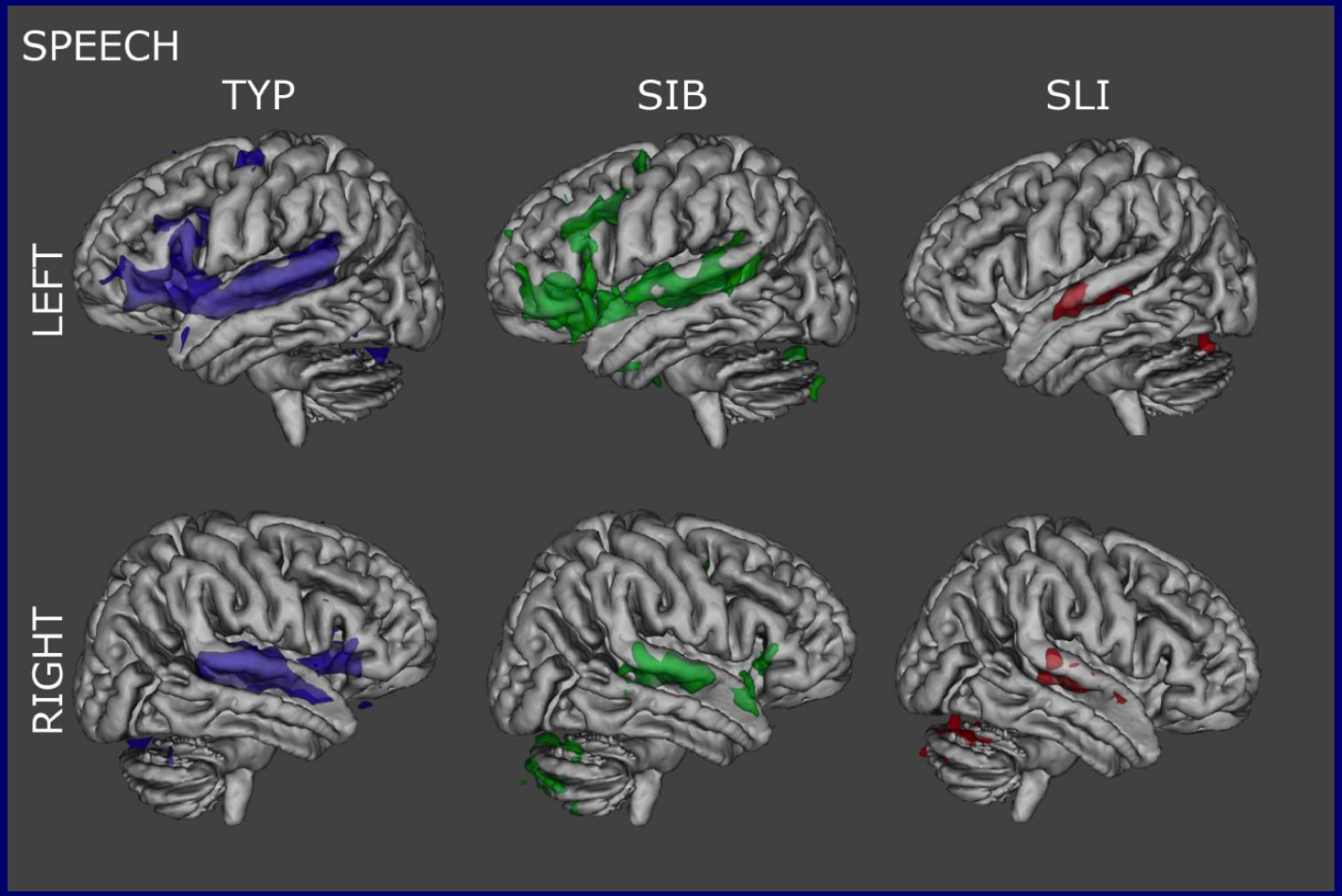
Badcock et al, 2012

Covert Naming Task



8 children with SLI; 6 unaffected siblings; 13 control children;

Activation to Covert Naming

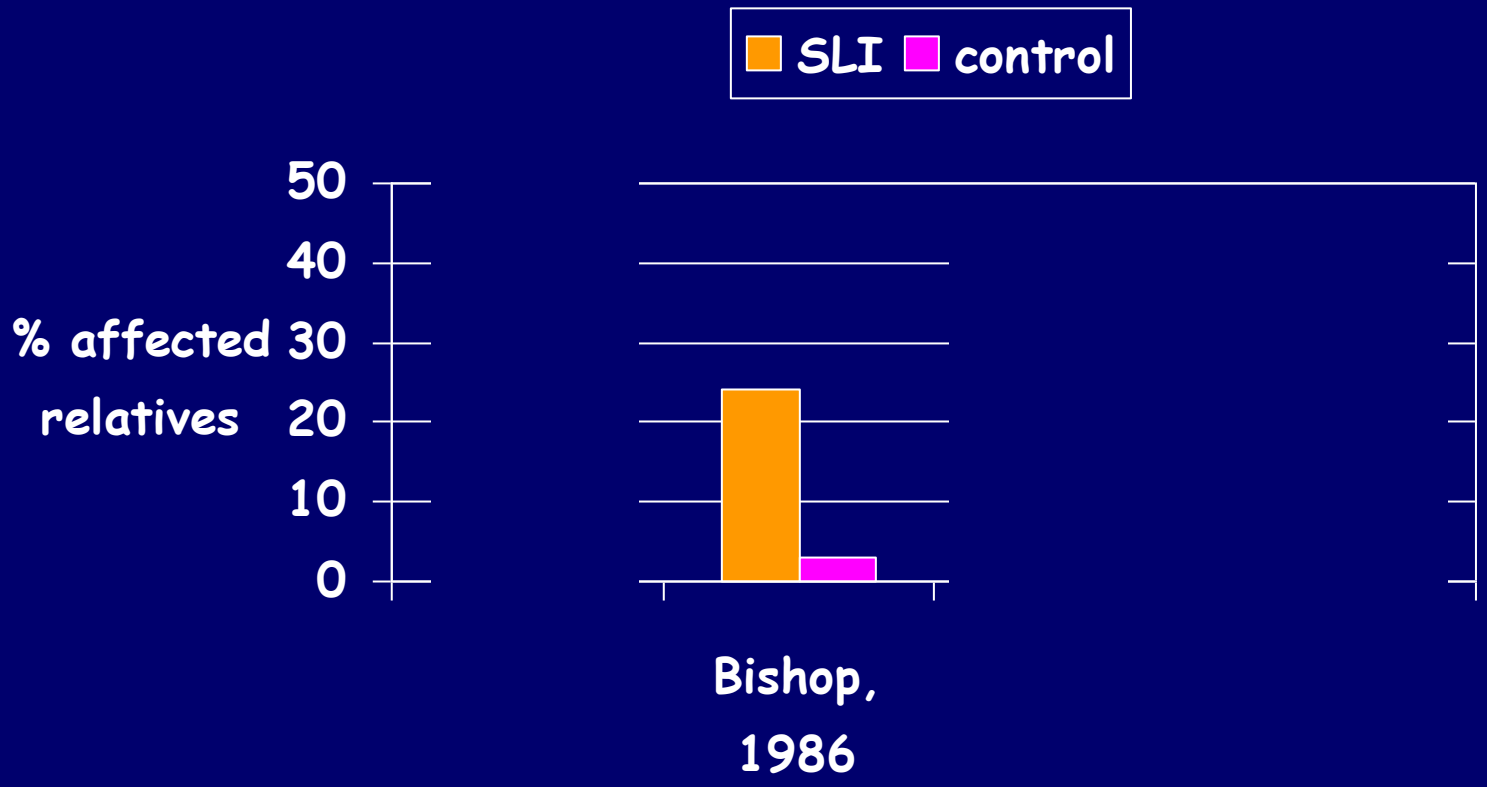


During covert auditory naming, SLI group show reduced activity in left IFG

Genetics

SLI: Family aggregation

Rates of language/learning difficulties higher in relatives of those with SLI, compared with controls



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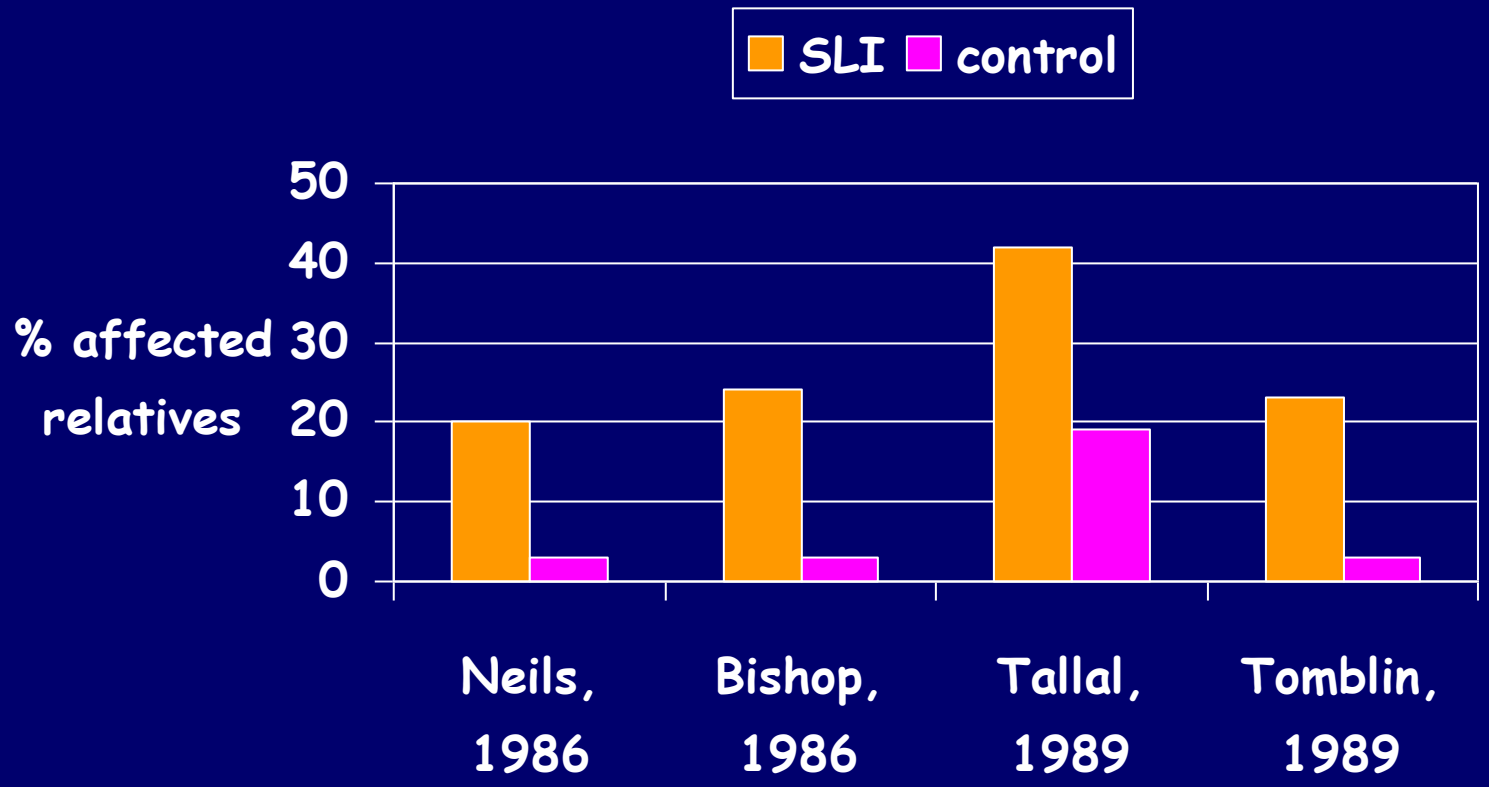


Twin Study Method

MZ twins: genetically identical

DZ twins: share 50% of polymorphic genes

Question:
Is concordance for disorder higher in MZ than in DZ twins?

Twin studies of SLI

	Probandwise concordance: same-sex twins	
	MZ	**DZ**
Lewis & Thompson, 1992	.86	.48
Bishop et al, 1995	.70	.46
Tomblin & Buckwalter, 1998	.96	.69

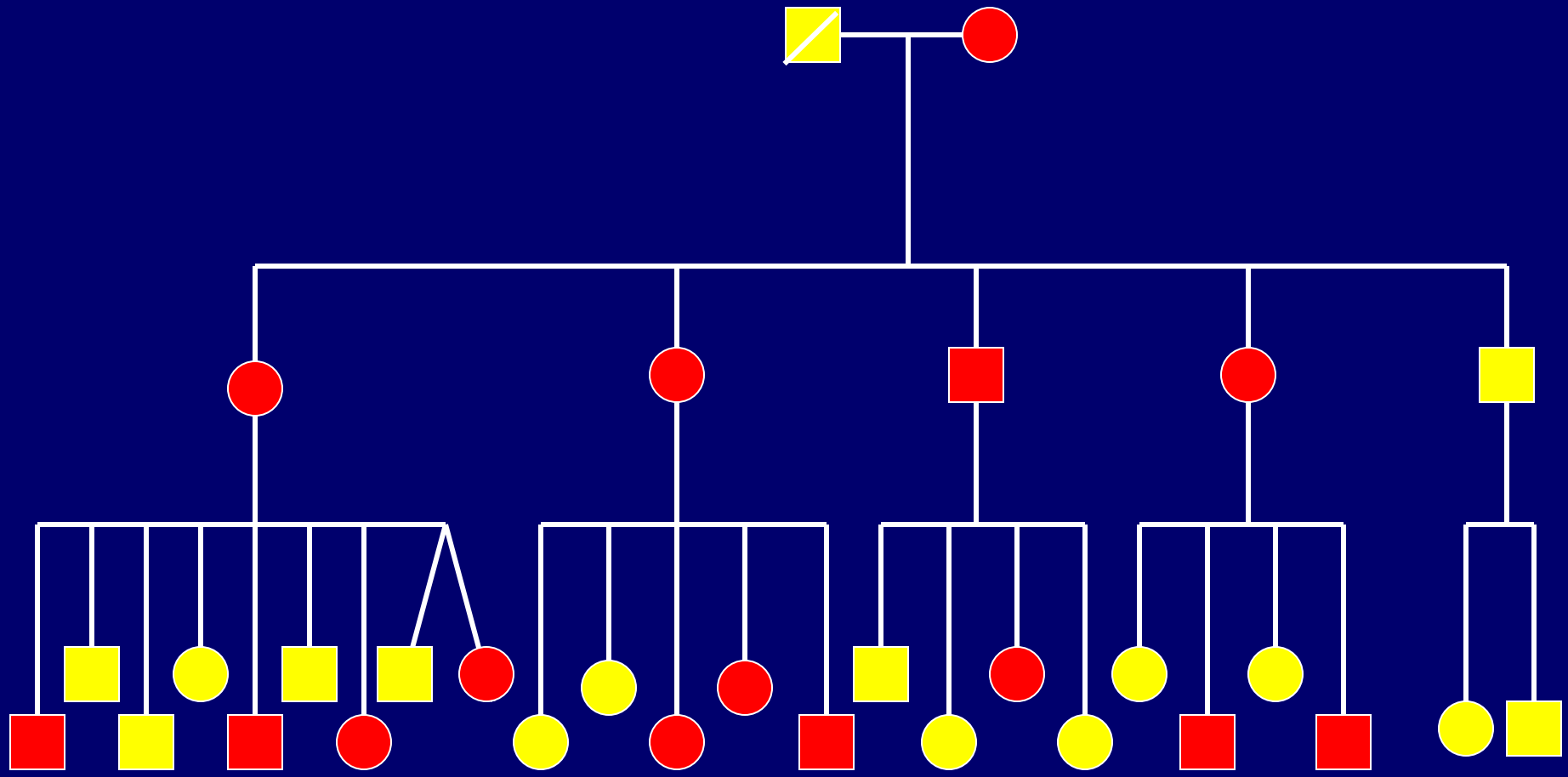
conclude……..

- Evidence for substantial genetic influence on SLI

- But lots of questions remain:

  - Which types of SLI are heritable, or not?
  - Can SLI be caused by a single gene?
  - Should we treat SLI as a single condition?
  - Is there genetic overlap between SLI and dyslexia and/or autism

Single gene disorder: KE family



Hurst et al, 1990; Gopnik, 1990;
Vargha-Khadem et al, 1995

KE family – a distinct type of SLI

- Distinctive clinical picture
  - Severe oromotor dyspraxia
  - Facial dysmorphology
  - Impairments of syntax
  - Poor at repeating nonsense words

- video uploaded to youtube by Stewart

Nature, October 4th 2001

A forkhead-domain gene is mutated in a severe speech and language disorder

Lai, C. S., Fisher, S. E., Hurst, J. A., Vargha-Khadem,F., & Monaco, A.

- FOXP2: gene on chromosome 7q31
- Single base mutation in affected individuals
- No allelic variation in people with normal language

Nature, August 22nd 2002

Molecular evolution of FOXP2, a gene involved in speech and language

Enard, W., Przeworski, M., Fisher, S. E., Lai, C. S. L. Wiebe, V., Kitano, T., Monaco, A. P., Paabo, S.

- FOXP2 differs in man and mouse in only 3 amino acid positions
- FoxP2 differs in man vs. chimp, gorilla, rhesus macaque in 2 amino acid positions
- i. e. 2/3 differences between human/mouse occurred on human lineage after separation from chimpanzee

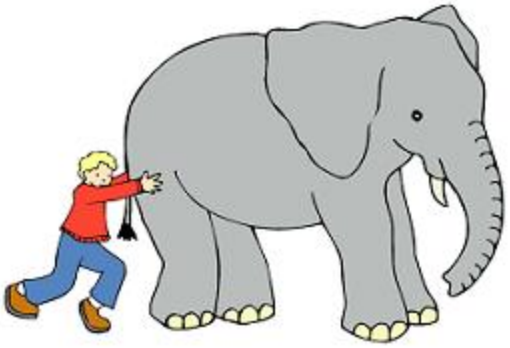
Is FOXP2 a 'grammar gene'?

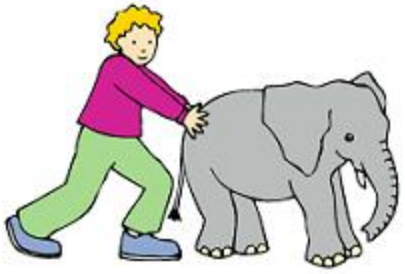
No!

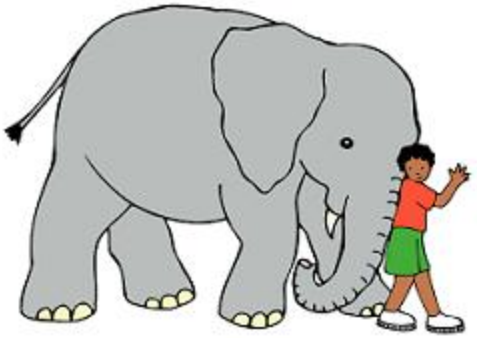
- FOXP2 is a transcription factor: regulates expression of many other genes, and expressed in multiple organs
- Affected members of KE family have problems beyond grammar
- Abnormality of subcortical regions involved in motor control

(Watkins et al, 2002)

Nevertheless,affected members poor at syntactic tasks, even when no speech is required, e.g.







The elephant pushing the boy is big

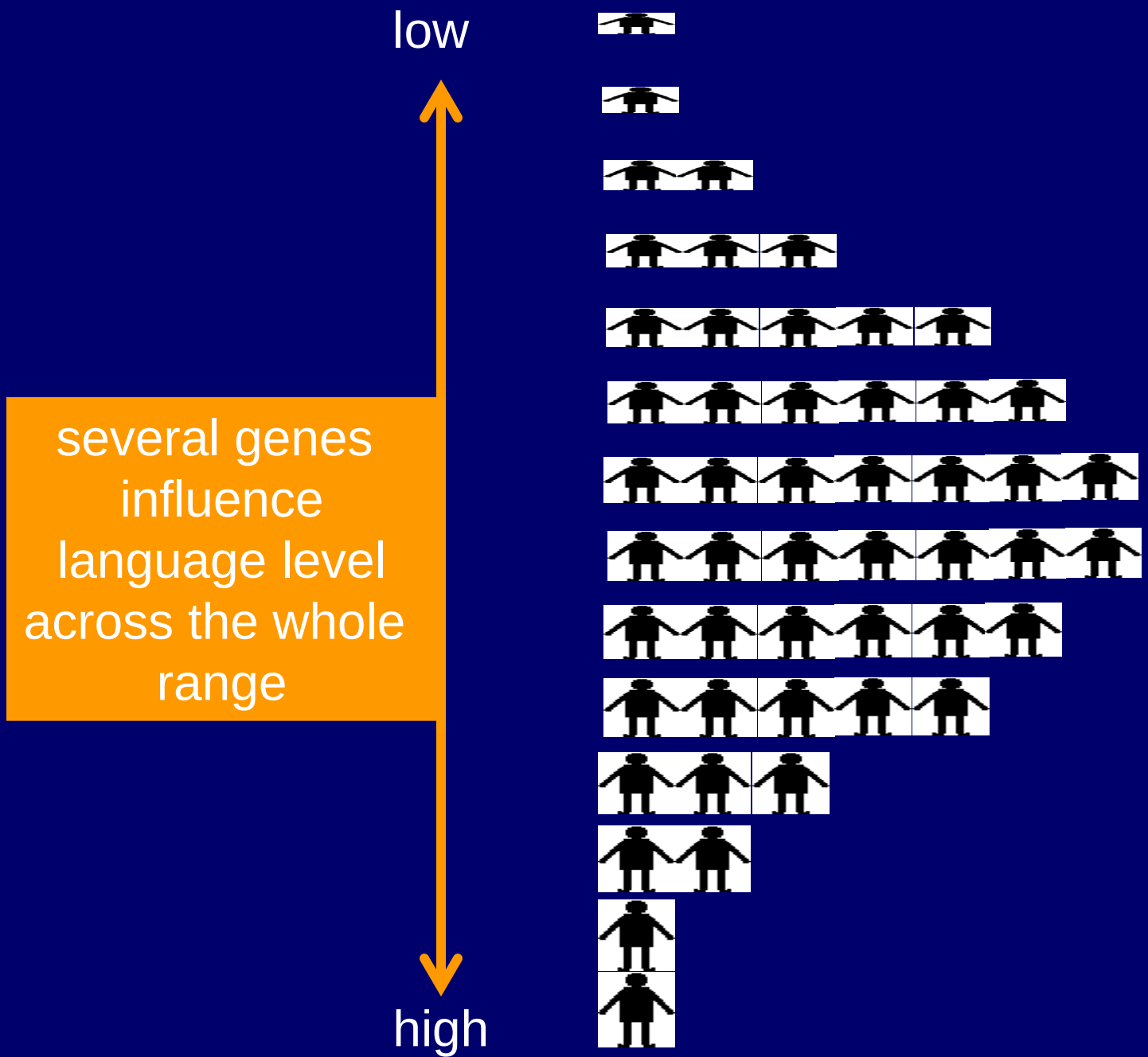
How to characterise FOXP2

- A transcription factor that affects expression of many other genes, and is expressed in multiple organs

- Human version of gene may be important for building brain areas involved in extracting hierarchical structure from linear input

Beyond FOXP2 - other cases of SLI

- No abnormality of FOXP2 seen in most cases of SLI
- Pedigree analysis does not show straightforward pattern suggestive of single gene disorder
- Different severity in MZ twins indicates role of environmental factors
- Finding genes will be harder: gene-disorder associations probabilistic


low
several genes influence language level across the whole range
high

How will we discover the genes?

- "Splitting" approach: look for genes related to component language skills that are affected in SLI

- "Lumping" approach: look for genes that might affect more than one disorder

Splitting

Move away from studying SLI to look at component skills

Some promising component skills

- Auditory processing skills (e.g., Tallal)

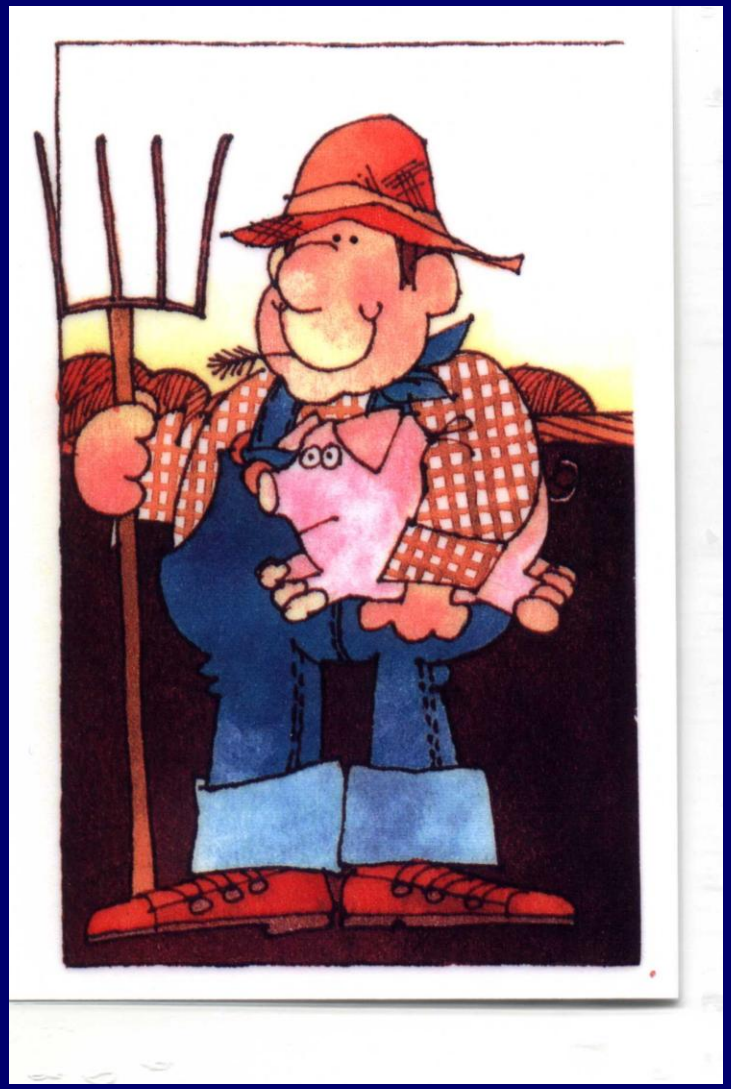
- Phonological short-term memory (e.g., Gathercole & Baddeley)

- Morphosyntax (e.g., Rice & Wexler)

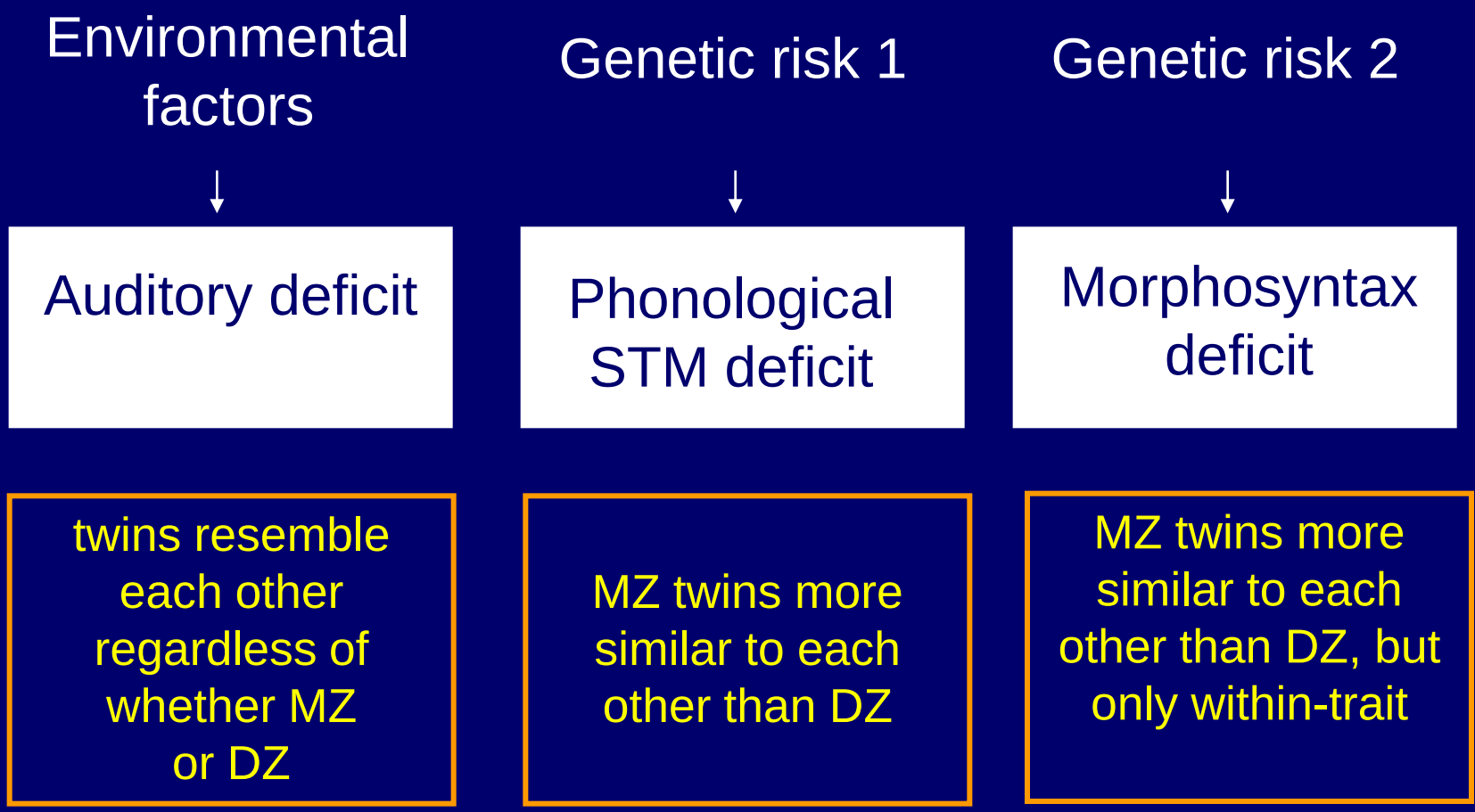
Rice-Wexler task

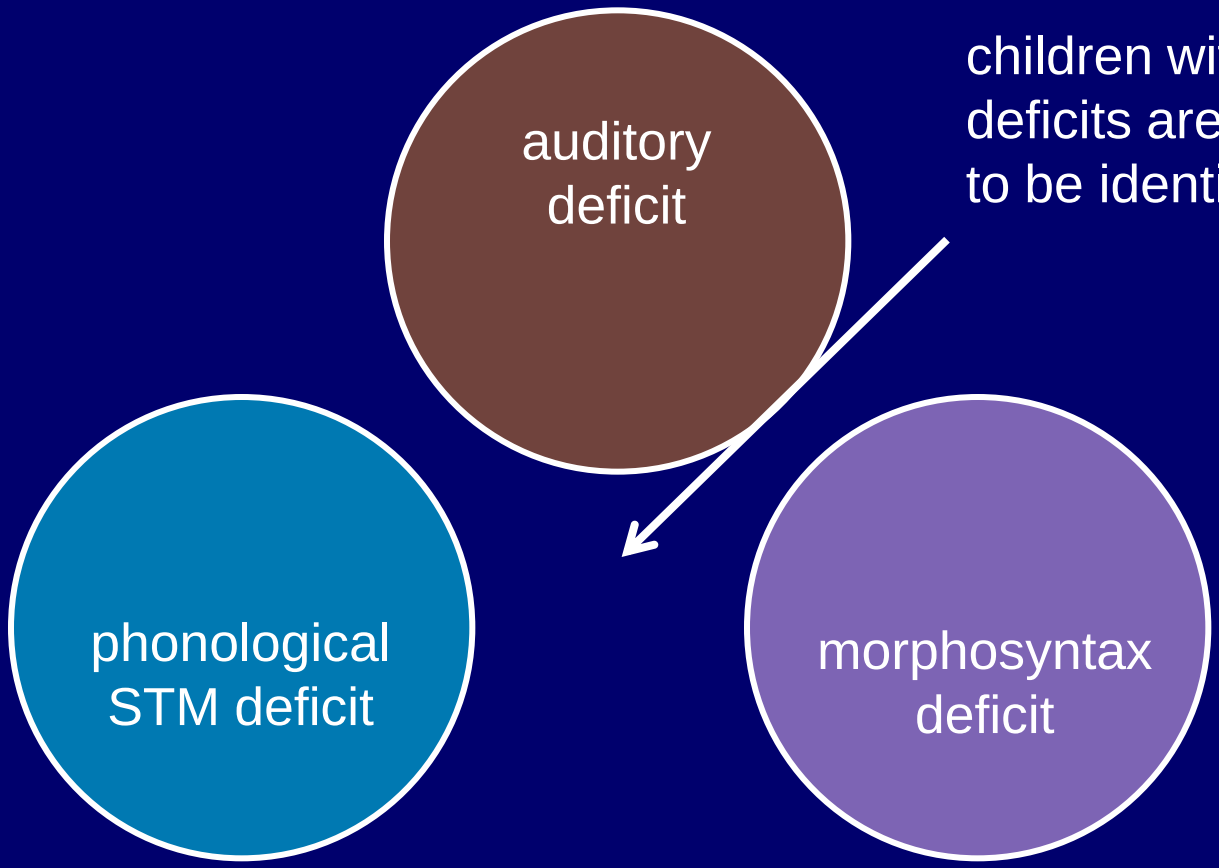
"Here the boy is raking; now he is done. Tell me what he did".

Here’s a farmer. Tell me what a farmer does.



Conclusions from twin studies





children with co-occurring deficits are most likely to be identified with SLI

Implications

- Different deficits may have different causes, but impact on language only severe when they co-occur

- Search for single cause explanations of SLI may be doomed - language is resilient enough to survive one deficit

Genetics: common misconceptions

- **Genes are the only thing that matter**

  NO! even in MZ twins, find different severity

- **No point in treating genetic disorders**

  NO! genetic analysis says nothing about effects of novel environmental experience

Genetic conditions that can be modified

- Hair colour!
- Diabetes
- Huntington’s disease

Dorothy Bishop
Oxford Study of Children's Communication Impairments,
Department of Experimental Psychology,
South Parks Road,
Oxford,
OX1 3UD,
England.

http://psyweb.psy.ox.ac.uk/oscci/