Family history interview of a broad phenotype in specific language impairment and matched controls

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Disclosure statement

I have no relevant financial or nonfinancial relationship(s) within the products or services described, reviewed, evaluated or compared in this presentation. /Nelli Kalnak
Heredity for language impairment - what do we know?

Prevalence rates of language problems in parents and siblings of:
- SLI probands 20-69%
- TD controls 6-7%

- Male > Female
- Severity = prevalence rates

- Severe impairment = parents more effective identifiers
WHY doing a family history study of specific language impairment?

Previous studies:

English speaking countries
Swedish sample

Mainly questionnaires
Family history interview

No broader phenotype
Broader phenotype
Participants with severe SLI (n=61)
School Language Units in the Stockholm Region

**Inclusion criteria**
- Specific Language Impairment at school admission
- Monolingual Swedish speaking
- 8-12 years old
- Normal hearing and sight

**Exclusion criteria**
- Adoption
Matched control children (n=100)
Swedish Birth Register

Matching criteria
- Birth year
- Gender
- Municipality of birth
- Country of origin of the mothers’
- District within municipality of birth

- Normal hearing
- Normal vision
- Monolingual
- Non adopted

No language related diagnoses or problem!
FAMILY HISTORY INTERVIEW

Consanguinity, deceased relatives, adoptions, abortions, miscarriages, IVF

Education
Occupation

Birth weight
Birth week
Postnatal status
Hearing & Vision
Language-Related Diagnoses and Problems (LRDPs)

GRANDPARENTS – PARENTS - SIBLINGS

DYSLEXIA
ADHD
AUTISM
ASPERGER
STUTTERING
CLEFT PALATE
MENTAL RETARDATION

LANGUAGE
READING
SCHOOL ACHIEVEMENT
ATTENTION/HYPERACTIVITY
SOCIAL COMMUNICATION
RESULTS
SLI probands’ pedigree 59 families

Control Children’s pedigree 100 families

Sum. n=415+59

n=733+100
SLI probands’
pedigree
59 families

Control Children’s
pedigree
100 families

% with any (one or more) LRDPs
Diagnoses OR problems?

**DIAGNOSES:**
- DYSLEXIA
- ADHD
- AUTISM
- ASPERGER
- STUTTERING
- CLEFT PALATE
- MENTAL RETARDATION

**PROBLEMS:**
- LANGUAGE
- READING
- SOCIAL COMMUNICATION
- SCHOOL ACHIEVEMENT
- ATTENTION/HYPERACTIVITY
<table>
<thead>
<tr>
<th>LANGUAGE-RELATED PROBLEMS</th>
<th>SLI-probands’ PARENTS N=118</th>
<th>Control children’s PARENTS N=200</th>
</tr>
</thead>
<tbody>
<tr>
<td>LITERACY</td>
<td>37.3%</td>
<td>5.0%</td>
</tr>
<tr>
<td>LANGUAGE</td>
<td>30.5%</td>
<td>1.5%</td>
</tr>
<tr>
<td>SOC.COMMUNICATION</td>
<td>16.9%</td>
<td>0%</td>
</tr>
<tr>
<td>SCHOOL ACHIEVEMENT</td>
<td>27.1%</td>
<td>1.5%</td>
</tr>
</tbody>
</table>
Co-occurrence

<table>
<thead>
<tr>
<th>Number of LRDPs</th>
<th>SLI parents (n=118)</th>
<th>Control parents (n=200)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>%</td>
<td>n</td>
</tr>
<tr>
<td>0</td>
<td>40</td>
<td>47</td>
</tr>
<tr>
<td>1</td>
<td>23</td>
<td>27</td>
</tr>
<tr>
<td>2</td>
<td>15</td>
<td>18</td>
</tr>
<tr>
<td>3</td>
<td>17</td>
<td>20</td>
</tr>
<tr>
<td>4</td>
<td>3</td>
<td>4</td>
</tr>
<tr>
<td>5</td>
<td>2</td>
<td>2</td>
</tr>
</tbody>
</table>
85% of the children with SLI
13% of the control children

... had parent(s) with LRDPs

Risk ratio 6.5
Clinical relevance

A family approach in clinical child language pathology is strongly advisable

• Parents own experience of LRDPs!
• Siblings – a risk group!
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Genes, Brain and Behavior
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Language-related **PROBLEMS**

**Language** Difficulty with language acquisition; late talkers (older than 3 years); having received speech/language therapy; diagnosis of language impairment.

**Reading** Reading impairment; difficulties in learning to read and write not due to either inadequate schooling or bilingualism; adults needing help in reading and understanding official letters; having a negative attitude towards reading; but not being diagnosed with dyslexia.
Language-related PROBLEMS

School achievement Difficulties in learning; having received special education at school; failing a school grade due to learning problems not related to e.g. social factors.

Attention/Hyperactivity Problems with attention and/or hyperactivity affecting learning or the social aspects of life; but not being diagnosed with ADHD.

Social communication Difficulties in making friends and/or maintaining friendships and relationships; speech anxiety; selective mutism.
PREVALENCE RATES of each language-related diagnoses or problem investigated

<table>
<thead>
<tr>
<th>LANGUAGE RELATED DIAGNOSIS AND PROBLEMS</th>
<th>Control Grandparents</th>
<th>SLI Grandparents</th>
<th>p^a</th>
<th>Control Parents</th>
<th>SLI Parents</th>
<th>p^a</th>
<th>Control Siblings</th>
<th>SLI Siblings</th>
<th>p^a</th>
</tr>
</thead>
<tbody>
<tr>
<td>LANGUAGE</td>
<td>n=397</td>
<td>n=220</td>
<td>.0040</td>
<td>n=200</td>
<td>1.5% (3)</td>
<td>30.5% (36)</td>
<td>&lt;.0001</td>
<td>n=136</td>
<td>8.0% (11)</td>
</tr>
<tr>
<td>READING</td>
<td>2.0% (8)</td>
<td>8.2% (18)</td>
<td>.0006</td>
<td>4.0% (8)</td>
<td>35.6% (42)</td>
<td>&lt;.0001</td>
<td>5.9% (8)</td>
<td>14.3% (11)</td>
<td>n.s.</td>
</tr>
<tr>
<td>SCHOOL ACHIEVEMENT</td>
<td>0</td>
<td>2.3% (5)</td>
<td>.0108</td>
<td>1.5% (3)</td>
<td>27.1% (32)</td>
<td>&lt;.0001</td>
<td>3.7% (5)</td>
<td>9.1% (7)</td>
<td>n.s.</td>
</tr>
<tr>
<td>SOCIAL COMMUNIC.</td>
<td>0</td>
<td>4.5% (10)</td>
<td>&lt;.0001</td>
<td>0</td>
<td>16.9% (20)</td>
<td>&lt;.0001</td>
<td>0.7% (1)</td>
<td>7.8% (6)</td>
<td>.0176</td>
</tr>
<tr>
<td>ATTENTION/HYPERACT</td>
<td>0.3% (1)</td>
<td>0.5% (1)</td>
<td>n.s.</td>
<td>0.5% (1)</td>
<td>8.5% (10)</td>
<td>.0006</td>
<td>2.2% (3)</td>
<td>2.6% (2)</td>
<td>n.s.</td>
</tr>
<tr>
<td>STUTTERING</td>
<td>0.8% (3)</td>
<td>0</td>
<td>n.s.</td>
<td>1.0% (2)</td>
<td>4.2% (5)</td>
<td>n.s.</td>
<td>0.7% (1)</td>
<td>0</td>
<td>n.s.</td>
</tr>
<tr>
<td>DYSLEXIA</td>
<td>0</td>
<td>0</td>
<td>n.s.</td>
<td>1.0% (2)</td>
<td>1.7% (2)</td>
<td>n.s.</td>
<td>2.2% (3)</td>
<td>9.1% (7)</td>
<td>n.s.</td>
</tr>
<tr>
<td>ADHD</td>
<td>0</td>
<td>0</td>
<td>n.s.</td>
<td>0.8% (1)</td>
<td>n.s.</td>
<td>1.5% (2)</td>
<td>5.2% (4)</td>
<td>n.s.</td>
<td>n.s.</td>
</tr>
<tr>
<td>CLEFT PALATE</td>
<td>0</td>
<td>0</td>
<td>n.s.</td>
<td>0.8% (1)</td>
<td>n.s.</td>
<td>1.5% (2)</td>
<td>1.3% (1)</td>
<td>n.s.</td>
<td>n.s.</td>
</tr>
<tr>
<td>AUSTISM/ASPERGER</td>
<td>0</td>
<td>0</td>
<td>n.s.</td>
<td>0</td>
<td>n.s.</td>
<td>0.7% (1)</td>
<td>1.3% (1)</td>
<td>n.s.</td>
<td>n.s.</td>
</tr>
<tr>
<td>MENTAL RETARDATION</td>
<td>0</td>
<td>0</td>
<td>n.s.</td>
<td>0</td>
<td>n.s.</td>
<td>0.7% (1)</td>
<td>1.3% (1)</td>
<td>n.s.</td>
<td>n.s.</td>
</tr>
<tr>
<td>Reading+Dyslexia</td>
<td>2.0% (8)</td>
<td>8.2% (18)</td>
<td>.0006</td>
<td>5.0% (10)</td>
<td>37.3% (44)</td>
<td>&lt;.0001</td>
<td>8.1% (11)</td>
<td>23.4% (18)</td>
<td>.0035</td>
</tr>
<tr>
<td>Soc.Comm.+Aut/Asp</td>
<td>0</td>
<td>4.5% (10)</td>
<td>&lt;.0001</td>
<td>0</td>
<td>16.9% (20)</td>
<td>&lt;.0001</td>
<td>2.2% (3)</td>
<td>9.1% (7)</td>
<td>n.s.</td>
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<tr>
<td>Att.Hyper.+ADHD</td>
<td>0.3% (1)</td>
<td>0.5% (1)</td>
<td>n.s.</td>
<td>0.5% (1)</td>
<td>9.3% (11)</td>
<td>.0002</td>
<td>3.7% (5)</td>
<td>7.8% (6)</td>
<td>n.s.</td>
</tr>
</tbody>
</table>
Specific Language Impairment (SLI)

Prevalence 5-7%

Comprehension
Production
Function

1-2% severe

Negative impact on social interaction and learning