Persistent Speech Sound Disorder (SSD) Outcomes in a Multigenerational Family

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Conflict of Interest Disclosure

We have no financial or non financial interest or related personal interest of bias in any organization whose products or services are described, reviewed, evaluated or compared in this presentation.

Presentation outline

1. Purpose of this study
2. Overview of existing research
3. Description of this study

Purpose

Describe an idiopathic speech sound disorder (SSD) phenotype in a large nuclear family (the PM Family)

Why study this family?

- High familial aggregation of SSD
- Distributional extremes
- Large family size
- Age > 9 years

Why study this family?

Behavioural studies: Suggest a strong genetic component
Molecular genetic studies: Mechanisms poorly understood, but

- families with many affected members of interest
- FOXP2 gene found in single extended pedigree (KE family)

Lewis et al., 2004; De Thorne et al., 2006. 
Lai et al., 2001
Vargha-Khadem et al., 1995
Why describe the phenotype?

- cognition
- language
- literacy
- speech
- numeracy
- phonological processing
- motor

Why describe the phenotype?

May help:
- understand heterogeneity
- cross-study comparisons
- facilitate research on genetic & neural correlates

Why study persistent SSD?

- Current research focus on early childhood SSD
- In persistent SSD research
  - variable phenotypic descriptions
  - more on known than unknown origin

Persistent Speech Sound Disorder

- Persistent: Speech errors > 8-9 years of age
- Speech Sound Disorder (SSD): Speech errors due to:
  structural, motor constraints, &/cognitive-linguistic constraints

Broad and Narrow definitions exist.

Prevalence of SSD unknown origin:

- 15.6% of 3-year-olds (Campbell et al., 2003)
- 3.8% of 6-year-olds (Shriberg et al., 2006)

Prevalence of Persistent SSD: known & unknown

- 3.6% of 8-year-olds (-1.2SD < mean) (Wren et al., 2012)
- 3.0% of 8-year-olds (-2SD < mean) (Wren et al., 2006)

Characteristics of persistent SSD?
Speech Characteristics

• Severity
  (Speake et al., 2012)

• CAS
  (Lewis et al., 2004a; Zaretsky et al., 2010)

• Dysarthria +/-
  (Fedorenko et al., 2015; Zaretsky et al., 2010)

• Orofacial apraxia
  (Vargha-Khadem, 1995)

Language & Literacy Characteristics

• Expressive language, literacy, & phonological processing impaired a

• Receptive language variable b

• Receptive language > expressive language trend

  a Lewis et al., 2004b; Speake et al., 2012; Zaretsky et al., 2010.
  b Lewis et al., 2004; Stackhouse, 1992.

Fine & Gross Limb Motor Characteristics

• Systematic assessment is rare

• Limb motor difficulties frequently been queried
  (Lewis et al., 2004b; Stackhouse & Snowling, 1992b; Zaretsky et al., 2010).

Educational/Vocational and Socio-emotional

• Little to no research specifically on persistent SSD

Study Participants

PM family: n=11

• 2 parents and 9 children
• 9 years to 55 years
• High aggregation of SSD (multiple-sound)

SSD History

<table>
<thead>
<tr>
<th>SSD History</th>
<th>Mum</th>
<th>Dad</th>
<th>Sib 1</th>
<th>Sib 2</th>
<th>Sib 3</th>
<th>Sib 4</th>
<th>Sib 5</th>
<th>Sib 6</th>
<th>Sib 7</th>
<th>Sib 8</th>
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</thead>
<tbody>
<tr>
<td>SSD Grouping</td>
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<tr>
<td>History of SSD</td>
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<td>Received therapy for SSD</td>
<td>Yes</td>
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<td>Limited preschool Rx</td>
<td>Yes</td>
<td>Yes</td>
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<tr>
<td>Previous CAS diagnosis</td>
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<tr>
<td>Intelligibility at 5 years</td>
<td>Poor</td>
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<td>Intelligibility at age 9 years</td>
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<tr>
<td>Rate of progress</td>
<td>Slow</td>
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</table>
Participant Groups

Criteria:
- the presence of multiple-sound SSD > 9 years
- the receipt of treatment for SSD > 9 years of age

Persistent SSD group: father & siblings 4, 5, 7, 8, 9 (n = 6)
Resolved SSD group: mother & siblings 1, 2, 3, 6 (n = 5)

Hypotheses:

(1) a core phenotype differentiated persistent from resolved SSD cases.
(2) the core phenotype resembled strongly familial persistent SSD cases in the literature.

Assessment Protocol:

<table>
<thead>
<tr>
<th>Domain</th>
<th>Measures</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cognition</td>
<td>WISC-IV, WAIS-III, spatial working memory</td>
</tr>
<tr>
<td>Language</td>
<td>CELF-4, PPVT-4</td>
</tr>
<tr>
<td>Literacy &amp; Numeracy</td>
<td>WIAT-II, WJ3</td>
</tr>
<tr>
<td>Speech</td>
<td>15 tasks - Madison Speech Assessment Protocol</td>
</tr>
<tr>
<td></td>
<td>Conversational speech</td>
</tr>
<tr>
<td>Phonological Processing</td>
<td>CTOPP Nonword Repetition subtest</td>
</tr>
<tr>
<td></td>
<td>Nonword Discrimination task</td>
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<tr>
<td></td>
<td>Lexical Discrimination task</td>
</tr>
<tr>
<td>Oro-motor</td>
<td>Structure, function, &amp; praxis tasks (MSAP)</td>
</tr>
<tr>
<td>Fine Motor</td>
<td>NEPSY-II: Finger Tapping, Imitating Hand</td>
</tr>
<tr>
<td></td>
<td>Positions, Manual Motor Seq. Body praxis task</td>
</tr>
</tbody>
</table>

Results

Case History & interview
- Developmental
- Academic
- Socio-emotional

Individual key results

Group comparisons

Results: Case History - Developmental

- Non syndromal
- No comorbid developmental diagnoses
- No childhood hearing impairment
- No medical history patterns
- Primary reason for referral = speech clarity

Case History: Academic

- All attended mainstream primary school
- Sibling 4 attended specialist high school
- Persistent cases: - Formal learning support
  - Trend to less years education
Case History: Socio-emotional

Lasting self-consciousness re speech: (All who had treatment)
Teasing and bullying related to speech: (All persistent SSD cases)
Psychological referral recommended: (1 Resolved & 4 Persistent cases)
Severe, persisting anxiety: (Sibs 4 & 9 Social Anxiety Disorder)

“My communication disorder has had a significant and profound impact on my life. Growing up I often felt left out because I wasn’t able to talk with other people, I wasn’t able to tell other people my thoughts or if I needed something.

It was heartbreaking because I knew what I wanted to say, but I couldn’t say it. I still feel deeply sad about not talking to others”

(Sibling 4 email using literacy support software; Carrigg et al., 2015 p. 46)
**Literacy and Numeracy: Persistent v Resolved SSD Groups**

<table>
<thead>
<tr>
<th>Measure</th>
<th>Persistent</th>
<th>Resolved</th>
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<tbody>
<tr>
<td></td>
<td>n</td>
<td>Mdn</td>
</tr>
<tr>
<td>Word Reading</td>
<td>5</td>
<td>58</td>
</tr>
<tr>
<td>Nonword reading</td>
<td>5</td>
<td>67</td>
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<tr>
<td>Word Spelling</td>
<td>6</td>
<td>62</td>
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<tr>
<td>Written Expression</td>
<td></td>
<td></td>
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<tr>
<td>- Holistic (0-4)</td>
<td>6</td>
<td>1</td>
</tr>
<tr>
<td>- Spelling (0-4)</td>
<td>6</td>
<td>0.00</td>
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<tr>
<td>- Punctuation (0-4)</td>
<td>6</td>
<td>0.50</td>
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<tr>
<td>Passage Comprehension</td>
<td>6</td>
<td>74</td>
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</tbody>
</table>

**Speech & Phonological Processing: Persistent v Resolved SSD Groups**

<table>
<thead>
<tr>
<th>Measure</th>
<th>Persistent</th>
<th>Resolved</th>
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<tbody>
<tr>
<td></td>
<td>n</td>
<td>Mdn</td>
</tr>
<tr>
<td>Nonword discrimination</td>
<td>6</td>
<td>71</td>
</tr>
<tr>
<td>Nonword repetition SS</td>
<td>5</td>
<td>1</td>
</tr>
<tr>
<td>Syllable repetition %</td>
<td>5</td>
<td>58</td>
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<tr>
<td>Lexical discrimination</td>
<td>6</td>
<td>93</td>
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<tr>
<td>Conversation PPC</td>
<td>5</td>
<td>86</td>
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<tr>
<td>Multisyllabic words PPC</td>
<td>6</td>
<td>60.5</td>
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<tr>
<td>Nonword repetition PPC</td>
<td>5</td>
<td>46</td>
</tr>
</tbody>
</table>

**Sibling 5, 17 years**

Discussion about his speech

“Longer words. Longer words have more syllables in it and, like, I have to get them together. Because it might got a /ch/ in the middle of the thing or a double ‘L’ word like loon, balloon, like a big word. The bigger the word, it’s harder”

“If I slow it down. But you can’t slow it down when you’re talking; you have to say it real fast”

**Sibling 5: 17 years. Multisyllabic Words Task (MSAP)**

- Emphasis
- Sympathise
- Fudgesicle
- Consciousness
- Fire extinguisher
- Statistician

“That’s hard, pass that one”

“It’s hard because I don't got someone talking saying it, like a computer saying, it’s hard”

**Father: Multisyllabic Words Task (MSAP)**

- Orchestra
- Specific
- Statistics
- Fire extinguisher
- Episcopal church

“I can’t say that one... I can’t”

**Oromotor Tasks**

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<th>St04</th>
<th>St05</th>
<th>St06</th>
<th>St07</th>
<th>St08</th>
<th>St09</th>
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<td>Oral Structure</td>
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<td>Orofacial Apraxia</td>
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<td>Oromotor function</td>
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<td>Speech-like task /s/</td>
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<td>Speech-like task DOK</td>
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</table>
### Motor: Persistent v Resolved Groups

<table>
<thead>
<tr>
<th></th>
<th>Persistent</th>
<th></th>
<th>Resolved</th>
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</thead>
<tbody>
<tr>
<td>Motor</td>
<td>n</td>
<td>Mdn</td>
<td>Range</td>
<td>n</td>
<td>Mdn</td>
<td>Range</td>
</tr>
<tr>
<td>Finger Tapping Repetition-SS</td>
<td>6</td>
<td>14</td>
<td>10-14</td>
<td>5</td>
<td>14</td>
<td>12-14</td>
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<tr>
<td>Finger Tapping Sequence-SS</td>
<td>6</td>
<td>10</td>
<td>8-11</td>
<td>5</td>
<td>11</td>
<td>5-12</td>
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<tr>
<td>Imitating Hand Positions SS</td>
<td>6</td>
<td>6</td>
<td>3-13</td>
<td>5</td>
<td>8</td>
<td>5-13</td>
</tr>
<tr>
<td>Manual Motor Sequences</td>
<td>6</td>
<td>2</td>
<td>1-4</td>
<td>5</td>
<td>4</td>
<td>1-4</td>
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</tbody>
</table>

### Summary

**Hypothesis 1: SUPPORTED**

- a core phenotype differentiated groups
- characterised multiple Verbal Trait Disorder

**Significant group differences (p < 0.01)**

1. Speech accuracy (multisyllabic, nonwords, conversation)
2. Verbal IQ
3. Receptive vocabulary
4. Expressive language
5. Written expression
6. Word reading & nonword reading
7. Word spelling
8. Nonword repetition
9. Nonword discrimination

**Persistent group characteristics: Core phenotype**

1. Current CAS (severe → mild)
2. Severe Expressive Language Disorder
3. Impaired single word receptive vocabulary
4. Receptive-Expressive language gap (RL > EL)
5. Lower verbal IQ than resolved cases
6. Impaired reading and spelling
7. Severely impaired phonological memory
8. Impaired nonword discrimination

**Resolved group characteristics**

1. Verbal IQ and nonverbal IQ: WNL
2. Expressive and receptive language: WNL
3. Speech: WNL → minimal distortion errors
4. Impaired nonword repetition

**Persistent group: Associated characteristics**

1. Academic difficulties: Formal learning support
2. Speech Intelligibility at 5 yrs: very poor
3. Speech Intelligibility at 9 years: fair → very poor
4. Progress rate: fair → very slow.

**Resolved group characteristics**

1. Verbal IQ and nonverbal IQ: WNL
2. Expressive and receptive language: WNL
3. Speech: WNL → minimal distortion errors
4. Impaired nonword repetition
Persistent group comparison to literature

Hypothesis 2: PARTIALLY SUPPORTED

- Caution required when comparing cases
- Phenotypic similarities and differences to KE family
- More similar to published idiopathic cases

Implications for management

multiple Verbal Trait Disorder

Assessment
- multiple domains
- challenges due to ↓ unintelligibility
- multidisciplinary
- family history

Thank you to...
The PM family, for their courage, generosity, and desire to help others with CAS.

- Sydney Children's Hospital Foundation Research Grant.
- Liz Kenway, clinical psychologist, for data collection assistance.
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