Psychology of Language

Prof. Jon Sprouse

01.24.13: Williams Syndrome and Specific Language Impairment
Is there a language faculty?

Do the processes that underly language form a separate cognitive faculty from the rest of cognition?

Or are those processes merely made possible by the power of our general cognitive abilities (e.g., general intelligence)?
The punchline: A double dissociation

Williams Syndrome and Specific Language Impairment form a double dissociation between intelligence and language ability.

**Williams Syndrome**
- general cognitive deficits
- no (or few) language deficits

**Specific Language Impairment**
- no general cognitive deficits
- language deficits
Williams Syndrome
The genetic cause

Williams Syndrome is caused by a deletion of about 20 genes on chromosome 7 - the specific location is called 7q11.23.

The number 7 refers to the chromosome.

The letter q refers to the long arm of the chromosome (the shorter arm is p).

The number 11 refers to a specific band that is visible on the chromosome when it is stained.

The number 23 refers to a sub-band of that primary band.

The exact role of many of these genes is still a matter for research. However ELN is the gene responsible for the protein elastin (plasticity in human organs), and LMK1 may be related to visual-spatial cognition.
Williams Syndrome leads to characteristic changes to facial features.

Even adults with WS tend to maintain a child-like (or “elf-like”) appearance.
Physiological Effects: Cardiovascular

The **elastin deficiency** caused by WS leads to a narrowing of the blood vessels (**stenosis**) throughout the body, most dangerously in the heart, lungs, and kidneys.

supravalvular aortic stenosis
Physiological Effects: the Brain

Williams Syndrome leads to a thickening of the cortex of the right hemisphere. But we have no idea how this would affect cognition.
Physiological Effects: the Brain

Williams Syndrome leads to increased folding (fissurization) of the brain. This is also called cortical complexity.
Physiological Effects: the Brain

Williams Syndrome also leads to an overall decrease in cortical volume. The decrease occurs in all lobes, and in both gray and white matter. However, the majority of the reduction is in white matter.
Physiological Effects: the Brain

Williams Syndrome also leads to an overall decrease in cortical volume. The decrease occurs in all lobes, and in both gray and white matter. However, the majority of the reduction is in white matter.
Williams Syndrome also leads to an overall decrease in cortical volume. The decrease occurs in all lobes, and in both gray and white matter. However, the majority of the reduction is in white matter.
Physiological Effects: the Brain

Williams Syndrome also leads to an overall decrease in cortical volume. The decrease occurs in all lobes, and in both gray and white matter. However, the majority of the reduction is in white matter.
Cognitive Effects: Intelligence

Williams Syndrome leads to a profound deficit in general intelligence as measured through standardized intelligence tests like the **Wechsler Adult Intelligence Scale**.

Intelligence scales are standardized such that 100 is mean for the population, with a standard deviation of 15.

This means that about 5% of people will have an IQ lower than 70. It also means that about 5% of the population will have an IQ above 130.
Cognitive Effects: Visual-Spatial

Model:

WS 11;1:

WS 9;1:

Control 6;1:
Cognitive Effects: Visual-Spatial

Model

Williams Age 11;1 KBIT 70 (RA)

Williams Age 11;1 KBIT 66 (BR)

Control Age 6;9 KBIT 116 (LC)
Cognitive Effects: Visual-Spatial

Model

Williams
Age 11;1
KBIT 70
(RA)

Williams
Age 11;1
KBIT 66
(BR)

Control
Age 6;9
KBIT 116
(LC)
Cognitive Effects: Visual-Spatial
Cognitive Effects: Visual-Spatial

Crucially these impairments cannot be explained by a deficit in visual-motor ability, as can be seen by success at tracing.

The deficit seems to be specific to visual-spatial tasks like drawing.
Cognitive Effects: Visual-Spatial

The specificity of the visual-spatial deficit can also be seen by comparing pure visual-spatial tasks like angle-identification, which WS patients fail at...
Cognitive Effects: Visual-Spatial

To distinct visual tasks like face recognition, which WS patients excel at.

(It should also be noted that there is a dedicated cortical area for human face recognition - the fusiform face area - which does not appear to be severely impaired in WS).
Comparison with Down Syndrome

Downs Syndrome is caused by a duplication of chromosome 21 - this leads to a total of 3 copies of this chromosome (we normally have 2), which is why it is also called trisomy 21.

DS results in similar performance on standardized IQ tests as WS.

Wechsler Intelligence Scale
Mean Score on WISC-R

WISC-R IQ

[Graph showing mean scores on the Wechsler Intelligence Scale]

- VIQ (Verbal)
- PIQ (Performance)
- FSIQ (Full Scale)

WMS N=10
DNS N=9
Comparison with Down Syndrome

Both WS and DS lead to visual-spatial deficits. However, they are distinct deficits: WS seems to preserve local details, but loses global details; DS seems to lose local details, but preserves global details.
Cognitive Effects: Language

One way in which Williams Syndrome appears to spare language ability, especially compared to Down Syndrome, is vocabulary.

Patients with Williams Syndrome have larger vocabularies, and are more likely to use it.
Cognitive Effects: Language

When asked to describe a picture, patients with WS will produce a longer, more coherent narrative, with far fewer grammatical mistakes.

Qualitative Examples of Increased Linguistic Evaluation in Adolescents with Williams Syndrome

WMS age 13
And he was looking for the frog. What do you know? The frog family! Two lovers. And they were looking. And then he was happy 'cause they had a big family. And said "good bye" and so did the frog. "Ribbit."

WMS age 17
Suddenly when they found the frogs... There was a whole family of frogs... And ah he was amazed! He looked... and he said "Wow, look at these... a female and a male frog and also lots of baby frogs". Then he take one of the little frogs home. So when the frog grow up, it will be his frog... The boy said "Good bye, Mrs. Frog... good bye many frogs. I might see you again if I come arounmd again". "Thank you Mr. Frog and Mrs. Frog for letting me have one of your baby frogs to remember him".

DNS age 13
There you are. Little frog. There another little frog. They in that... water thing. That's it. Frog right there.

DNS age 18
Thy're hiding; see the frogs... the baby frogs. Uh, the boy, and, and the dog saw the frogs. The frog's got babies. The boy saw the... no, the boy say good bye.

(M. Mayer, "Frog Where are You")

(Reilly, Klima & Bellugi, 1990)
Cognitive Effects: Language

When asked to describe a picture, patients with WS will produce a longer, more coherent narrative, with far fewer grammatical mistakes.

WMS age 17, Full Scale IQ = 50

Once upon a time when it was dark at night...the boy had a frog. The boy was looking at the frog...sitting on the chair, on the table, and the dog was looking through...looking up to the frog in a jar. That night he slept and slept for a long time, the dog did. But, the frog was not gonna go to sleep. And when the frog went out...the boy and the dog were still sleeping. The next morning it was beautiful in the morning. It was bright and the sun was nice and warm. Then suddenly when he opened his eyes...he looked at the jar and then suddenly the frog was not there. The jar was empty. There was no frog to found (whispered).

DNS age 18, Full Scale IQ = 55

The frog is in the jar. The jar is on the floor. The jar on the floor. That's it. The stool is broke. The clothes is laying there.
Cognitive Effects: Language

The contrast between visual-spatial and language abilities in WS is particularly striking when patients are asked to describe the pictures that they draw:

FIG. 2.6. Contrast between visuospatial and language abilities in WS. (a) Drawing of an elephant by an 18-year-old WS woman, whose IQ is 49. (b) Her verbal description of an elephant.

And what an elephant is, it is one of the animals. And what the elephant does, it lives in the jungle. It can also live in the zoo. And what it has, it has long gray ears, fan ears, ears that can blow in the wind. It has a long trunk that can pick up grass, or pick up hay....If they're in a bad mood it can be terrible...If the elephant gets mad it could stomp; it could charge, like a bull can charge. They have long big tusks. They can damage a car...It could be dangerous. When they're in a pinch, when they're in a bad mood it can be terrible. You don't want an elephant as a pet. You want a cat or a dog or a bird...
Given an appropriate context, patients with WS will spontaneously produce complex grammatical constructions.
Cognitive Effects: Language

Given an appropriate context, patients with WS will spontaneously produce complex grammatical constructions.
Cognitive Effects: Language

Given an appropriate context, patients with WS will spontaneously produce complex grammatical constructions.

This is an object relative clause, which we have already seen is relatively difficult to process (and causes increased activation in LiFG).

Which cow is Max (the rat) looking at?

“The cow who um the boy is pointing at.”

12 year old, Williams Syndrome, IQ = 56
Specific Language Impairment
What is Specific Language Impairment?

Specific Language Impairment (SLI) is a developmental disorder that specifically affects language, without any other disorder that can explain it (hearing, general cognitive development, etc).

General clinical symptoms:

Production delay in first words

Deviant production of speech sounds

Simplified grammatical productions (omission of tense markers, etc)

Restricted vocabulary in both production and comprehension

Trouble repeating words or sentences (perhaps due to short term memory deficits)

Comprehension difficulty with complex sentences and/or rapid speech
How is SLI diagnosed?

By definition, SLI is a deficit in language development without any other accompanying cognitive or sensory deficits that could explain it. This means that a diagnose of SLI requires the elimination of any other possible causes.

**General Diagnostic criteria:**

Language production and/or comprehension in lowest 10% for age on standardized test

Nonverbal IQ and other cognitive abilities fall within normal limits for age

No hearing loss, physical abnormality of the speech organs, or brain damage

No deprivation of language input in the environment

The rate of SLI in kindergarten-aged children has been estimated to be **as high as 7%**; however, such estimates are likely inflated, as large scale studies have not combined both inclusionary criteria (language impairment) and exclusionary criteria (nonverbal cognitive abilities).
What causes SLI?

The cause of Specific Language Impairment (SLI) is likely genetic; however, unlike Williams Syndrome, the genetic cause has not been identified.

So how do we know it is genetic?

The primary evidence comes from the rate of incidence between different types of twins.

The idea is that siblings are generally exposed to the same environmental factors (parenting, education, nutrition, etc) but can vary in genetic relatedness.

The proportion of pairs of monozygotic twins (one egg - identical twins) with SLI is much higher than the proportion of pairs of dizygotic twins (two eggs - fraternal twins).

Furthermore, in cases where only one member of monozygotic twins has SLI, the other tends to show some language impairment, though perhaps not severe enough to meet the diagnostic criteria for SLI.
Theories of SLI: Functional Morphology

One prominent theory of SLI is that individuals with SLI have a deficit in the use of functional morphology: the past tense (-ed), the plural (-s), comparatives (-er), etc.

| Table 2. Ability to mark novel words grammatically (% correct) |
|---|---|---|---|
| Grammar | Language | Controls | Impaired |
| Past tense | English (in England) | 95.4 | 38.0 |
| | English (in Canada) | 93.5 | 52.3 |
| | Greek | 87.1 | 20.0 |
| | French | 92.6 | 33.3 |
| | Japanese | 89.1 | 37.0 |
| Plurals | English (in England) | 95.7 | 57.0 |
| | English (in Canada) | 99.2 | 58.3 |
| | Greek | 79.8 | 42.1 |
| Comparatives | English (in England) | 74 | 21 |
| Compounds | Japanese | 80.5 | 20.2 |
| | Greek | 93.6 | 12.8 |
| Diminutives | Greek | 83.9 | 40.2 |

In each of these tests the subjects were given a context which required that a grammatical rule be applied to a novel word: This pencil is weff. This pencil is even...
Theories of SLI: Functional Morphology

The primary problem with the functional morphology theory is that the mistakes that children with SLI make are **errors of omission**:

Yesterday, I walk.

They **never misuse morphology**:

Yesterday, I walk **er**.  Yesterday, I walks.

<table>
<thead>
<tr>
<th>Table 1. Ability to produce tense marking (% correct)</th>
</tr>
</thead>
<tbody>
<tr>
<td>% correct</td>
</tr>
<tr>
<td>----------</td>
</tr>
<tr>
<td>Impaired</td>
</tr>
<tr>
<td>Controls</td>
</tr>
</tbody>
</table>

*Subjects were given items like: Everyday I walk to school. Just like everyday, yesterday I.... This task requires the subject to recognize that the temporal context specified in the second sentence requires a particular verb form.*
Theories of SLI: Optional Infinitives

The optional infinitives theory attempts to account for the fact that children with SLI never misuse functional morphology, they only omit it.

Typically developing children go through a stage where they also omit functional morphology. Wexler (1990) calls this the optional infinitive stage because infinitives are a grammatical option for these children (even though they aren’t an option for adults):

**Adult grammar:** * Daddy walk to work.

**Child grammar:** Daddy walk to work.

The optional infinitive approach to SLI claims that children with SLI simply remain in the optional infinitive stage longer than typically developing children, and perhaps remain there indefinitely (if the SLI doesn’t resolve with age).

<table>
<thead>
<tr>
<th>age group</th>
<th>% OIs</th>
</tr>
</thead>
<tbody>
<tr>
<td>1:07-2:00</td>
<td>83% (126/152)</td>
</tr>
<tr>
<td>2:01-2:06</td>
<td>64% (126/198)</td>
</tr>
<tr>
<td>2:07-3:00</td>
<td>23% (57/253)</td>
</tr>
<tr>
<td>3:01-3:07</td>
<td>7% (29/415)</td>
</tr>
</tbody>
</table>
Theories of SLI: Auditory Processing

One problem with the optional infinitive approach is that children with SLI tend to have hearing deficits beyond functional morphology.

Although children with SLI perform well on hearing tests, the tones in those tests tend to be relatively long and static:

However, on discrimination tasks where two tones are presented in quick succession, children with SLI require much longer pauses between the tones (called the inter-stimulus interval, or ISI) for successful discrimination:
Theories of SLI: Auditory Processing

The fact that children with SLI have difficulty discriminating sounds in quick succession may explain difficulties with language acquisition.

Functional morphemes are very short in duration, and occur in quick succession with the root word:

```
walk ------ s
```

One potential consequence is that children with SLI may have difficulty learning morphological paradigms, and instead simply persist in the infinitive stage.
A specific type of SLI: the KE family

There is a family in London that exhibits a particularly severe form of SLI.

What is particularly interesting about this family is that the deficit has appeared in nearly half of the family members, across at least three generations. This has allowed researchers to investigate both the **behavioral deficits** and any **genetic differences between family members**.

Genetic tests have revealed a mutation in the **FOXP2** gene, which is located on chromosome 7, specifically at 7q31.
A specific type of SLI: the KE family

Comparisons of unaffected and affected family members on a wide battery of tests reveals that the deficits in the KE family are much broader than the deficits reported in the general SLI population:

Affected members show the typical **functional morphology** deficits:

But they also show deficits in **oral-facial** abilities:

---

**Fig. 2.** Production of tenses. Scores are means ± standard errors. See Table 2 for examples of test items.

**Fig. 3.** Imitation of oral and facial movements. Scores are means ± standard errors.
A specific type of SLI: the KE family

Affected members also show language deficits beyond the typical functional morphology deficits:

<table>
<thead>
<tr>
<th>Test (ref.) [Instructions]</th>
<th>Maximum score</th>
<th>Score</th>
<th>t value</th>
<th>P</th>
<th>P (third generation only)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Digit span (9, 10) [Repeat this list of numbers (forwards and backwards)]</td>
<td>10 ± 3</td>
<td>6.3 ± 2.40</td>
<td>10.00 ± 2.83</td>
<td>3.14</td>
<td>0.005</td>
</tr>
<tr>
<td>Alphabet words [Repeat this word (each begins with a different letter)]</td>
<td>39</td>
<td>29.58 ± 4.66</td>
<td>38.75 ± 0.71</td>
<td>6.70</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Repetition of words (11) [Repeat this word]</td>
<td>40</td>
<td>18.00 ± 5.92</td>
<td>37.33 ± 2.81</td>
<td>7.50</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Repetition of nonwords (11) [Repeat this nonword exactly as I say it]</td>
<td>40</td>
<td>16.38 ± 5.44</td>
<td>34.88 ± 5.38</td>
<td>7.59</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Lexical decision (11) [Is this a real English word?]</td>
<td>60</td>
<td>46.91 ± 6.95</td>
<td>54.57 ± 4.89</td>
<td>2.53</td>
<td>0.022</td>
</tr>
<tr>
<td>Sentence repetition (12) [Repeat this sentence exactly as I say it]</td>
<td>20</td>
<td>3.64 ± 5.01</td>
<td>12.25 ± 5.75</td>
<td>3.48</td>
<td>0.003</td>
</tr>
<tr>
<td>Object naming* (13) [Tell me the name of the object in this picture]</td>
<td>36</td>
<td>26.33 ± 4.38</td>
<td>30.13 ± 2.80</td>
<td>0.02</td>
<td>0.903</td>
</tr>
<tr>
<td>Picture vocabulary* (14) [Show me the picture for this word]</td>
<td>100 ± 15</td>
<td>65.38 ± 11.37</td>
<td>85.13 ± 10.84</td>
<td>4.26</td>
<td>0.054</td>
</tr>
<tr>
<td>Phoneme deletion (15) [Say this nonword without its first sound—e.g., varg → arg]</td>
<td>24</td>
<td>12.50 ± 5.62</td>
<td>22.14 ± 1.57</td>
<td>5.58</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Phoneme addition (15) [Say this nonword adding this first sound—e.g., arg → varg]</td>
<td>24</td>
<td>14.08 ± 5.98</td>
<td>21.00 ± 3.65</td>
<td>2.76</td>
<td>0.013</td>
</tr>
<tr>
<td>Nonword reading (15) [Read this nonword (pronounceable but meaningless)]</td>
<td>30</td>
<td>9.08 ± 5.11</td>
<td>23.00 ± 9.76</td>
<td>4.12</td>
<td>0.001</td>
</tr>
<tr>
<td>Nonword spelling (15) [Write this nonword as if it were a real English word]</td>
<td>30</td>
<td>7.83 ± 7.30</td>
<td>19.86 ± 7.95</td>
<td>3.36</td>
<td>0.004</td>
</tr>
<tr>
<td>Rhyme production [Tell me a word that rhymes with this word]</td>
<td>24</td>
<td>13.00 ± 5.73</td>
<td>20.86 ± 7.47</td>
<td>2.52</td>
<td>0.023</td>
</tr>
</tbody>
</table>

Scores are presented as mean ± SD.

*Scores on this test correlated significantly with performance IQ (P < 0.05); the values reported are therefore based on an analysis of covariance.
The punchline: A double dissociation

Williams Syndrome and Specific Language Impairment form a double dissociation between intelligence and language ability.

**Williams Syndrome**
- general cognitive deficits
- no (or few) language deficits

**Specific Language Impairment**
- no general cognitive deficits
- language deficits