LING 1010

Language and Mind

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03.25.19: Williams Syndrome and Specific Language Impairment
An important double dissociation

Williams Syndrome and Specific Language Impairment form a double dissociation between intelligence and language ability. This suggests that language ability is independent of general of intelligence.

**Williams Syndrome**
- Affects general intelligence
- Has no (or few) effects on language

**Specific Language Impairment**
- Has no effects on general intelligence
- Affects language acquisition
What is a double dissociation?

Double dissociation is just a fancy way of saying that two abilities can vary independently of each other.

When we say that language ability and intelligence are doubly dissociated, what we are really saying is that all four combinations of ability are possible:

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<thead>
<tr>
<th>Intelligence</th>
<th>Language</th>
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<tr>
<td>good</td>
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<tr>
<td>bad</td>
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Double dissociations are important tools in cognitive science because they show us that the two abilities are independent of each other. There is no necessary relationship between them.
Williams Syndrome:

Cognitive and Visual deficits, but no (or very few) language deficits
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.

The constellation of features is often described as youthful, even for adults.
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.
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 typically developing adults will have an IQ lower than 70. It also means that about 5% of typically developing adults will have an IQ above 130.
Individuals with WS tend to have trouble with visual-spatial processing, which can be seen when they attempt to recreate shapes or pictures by drawing. Here you can see older children with WS are worse at drawing shapes than a younger child without WS.
Cognitive Effects: Visual-Spatial

Here you can see individuals with WS do notice that there is fine-grained detail inside of the circles, but they have trouble organizing that detail correctly.
Cognitive Effects: Visual-Spatial

Here is another example of individuals with WS noticing the fine-grained detail, but having trouble organizing that detail into a coherent whole.
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.

This shows that they can perceive the shapes accurately, and can even create motor commands to create them.

The deficit seems to be specific to visual-spatial tasks that require independent recreation.
Comparing Downs Syndrome and Williams Syndrome:

Both show general intelligence and visual-spatial deficits, but WS shows far fewer language deficits.

(This makes it clear that general intelligence can be affected without affecting language.)
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 (2 is typical), which is why it is also called trisomy 21.

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

[Graph showing Wechsler Intelligence Scale Mean Score on WISC-R]
Comparison with Down Syndrome

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

If one were to look at DS alone, one might conclude that general intelligence deficits and language deficits go hand-in-hand. But a comparison with WS shows that general intelligence deficits can occur without language deficits.

Patients with Williams Syndrome tend to have larger productive vocabularies (I am not sure about receptive vocabulary differences — the studies that I have seen have focused on productive vocabulary).
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.
Specific Language Impairment:

Language deficits, but no cognitive deficits
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 diagnosis 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.
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 SLI language deficits:

But they also show deficits in oral-facial abilities:

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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.
Some conclusions: Double Dissociation

Williams Syndrome and Specific Language Impairment form a double dissociation between intelligence and language ability. This suggests that language ability is independent of general intelligence.

**Williams Syndrome**
- Affects general intelligence
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In short, there appears to be a biological basis for investigating the language faculty as a distinct system from general intelligence.