LING 1010

Language and Mind

Prof. Jon Sprouse

03.17.21: Williams Syndrome and Specific Language Impairment

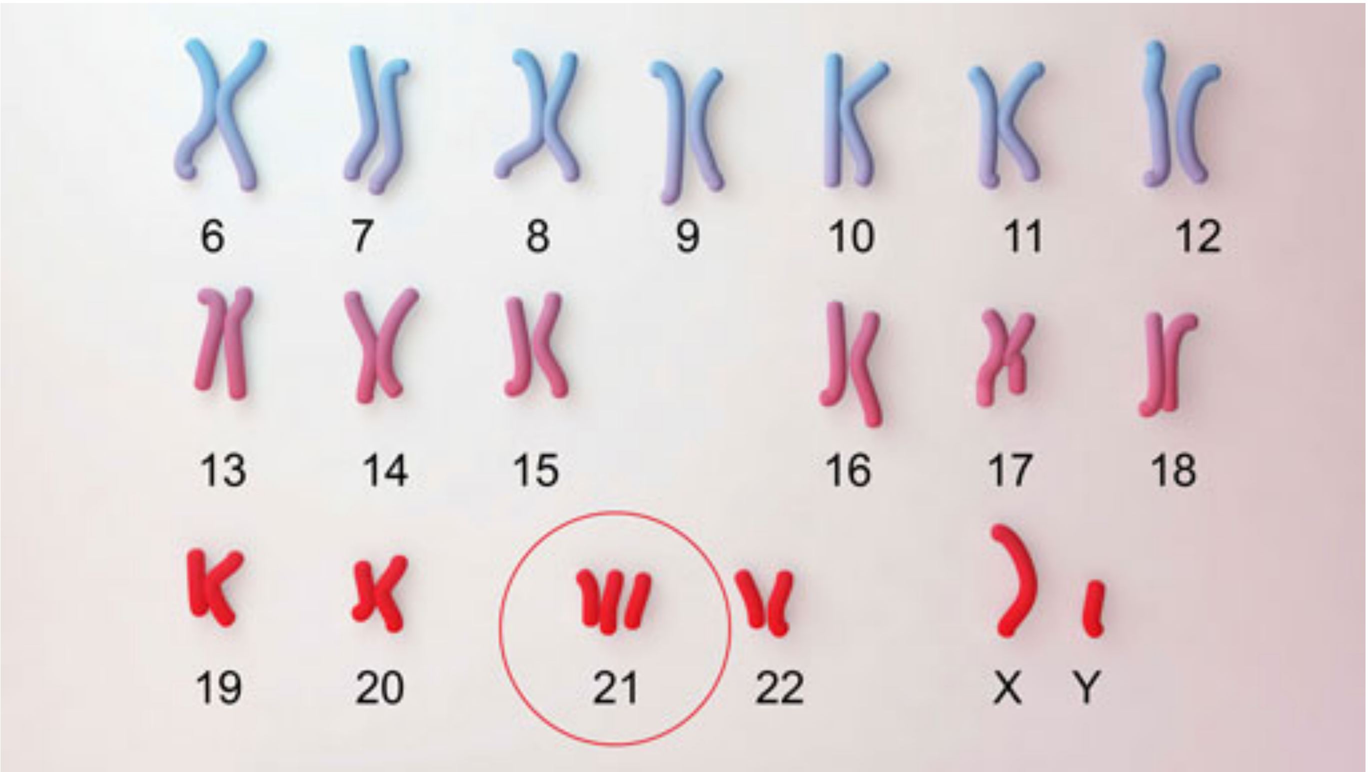
Big Question: Maybe it is all just intelligence?

Prediction: As intelligence varies, so will language ability

A misleading correlation:
Down Syndrome (Trisomy 21)

Down Syndrome/Trisomy 21

Down Syndrome is a genetic disorder caused when abnormal cell division results in an extra full or partial copy of chromosome 21. [Mayo Clinic]

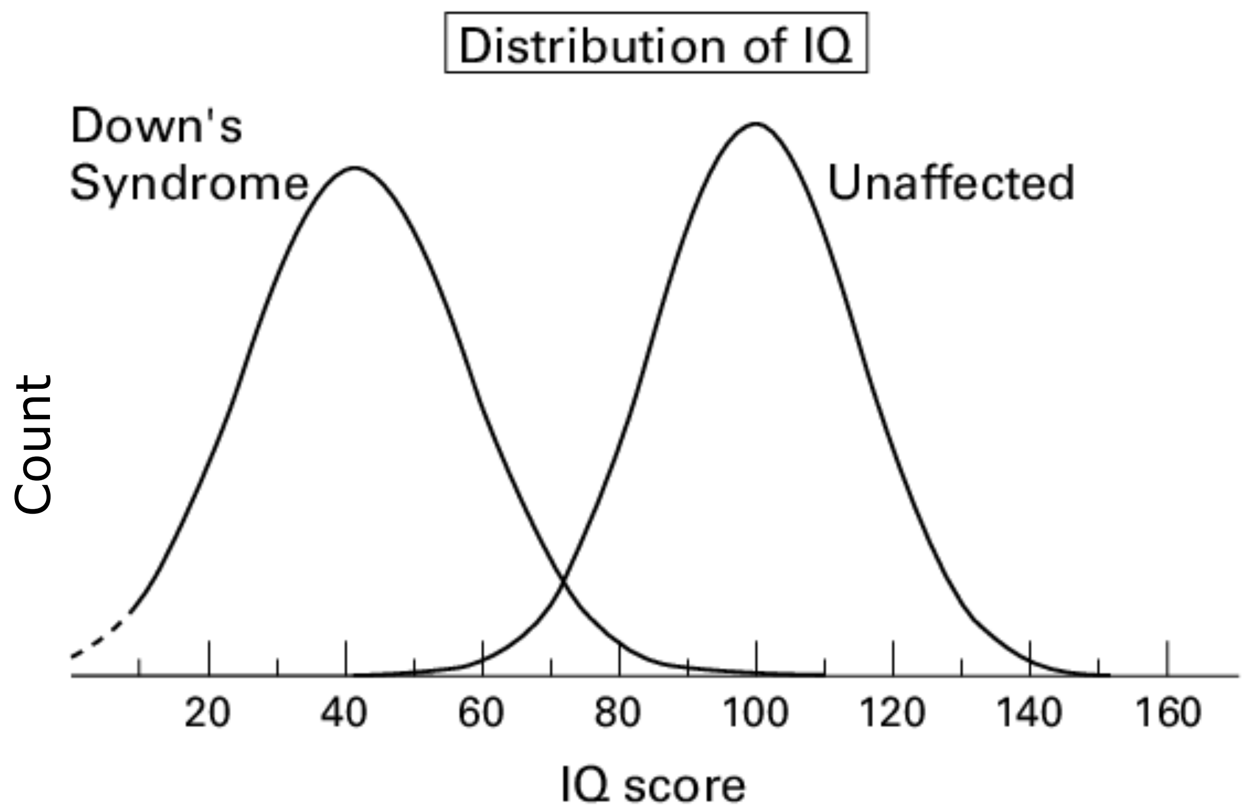


Cognitive Effects: Intelligence

Down 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.



Language effects

Down Syndrome often results in noticeable effects on language ability. Here you can see the responses of two children asked to describe a page from a picture book:

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

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.

(Reilly, Klima & Bellugi, 1990)

The plan for today

Down Syndrome/Trisomy 21 is relatively common compared to other genetic developmental disorders, so the fact that it appears to cause effects on <u>both</u> general intelligence and language can give the impression that the two are linked.

So, today, we will look at two less common developmental disorders that show that this correlation is <u>not absolute</u> (that is, not necessary): intelligence and language can vary **independently of each other.**

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:

	Intelligence	**Language**
option 1:	unaffected	unaffected
option 2:	affected	affected
option 3:	unaffected	affected
option 4:	affected	unaffected

Under a double dissociation, all four options are possible!

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

The genetic cause
(is very different than Down Syndrome)

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

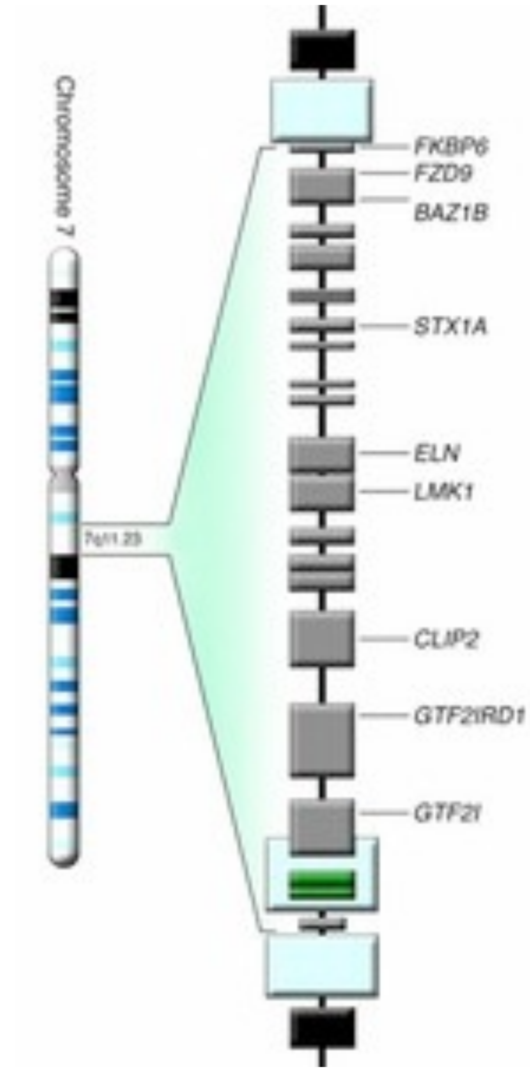
The number 7 refers to the chromosome.

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.

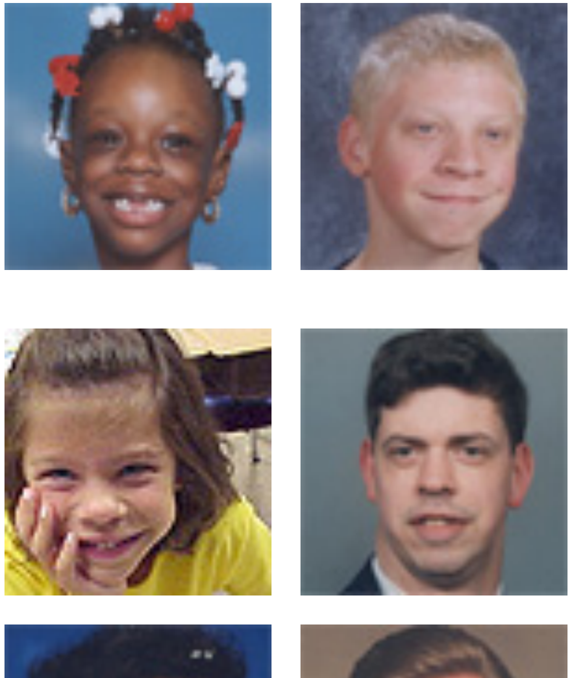
The exact role of many of these genes is still a matter for research. However **ELN** is the gene responsible for the protein **elastin** (plasticity in human organs), and **LMK1** may be related to visual-spatial cognition.



Physiological Effects: Facial Features

Williams Syndrome leads to characteristic changes to facial features.

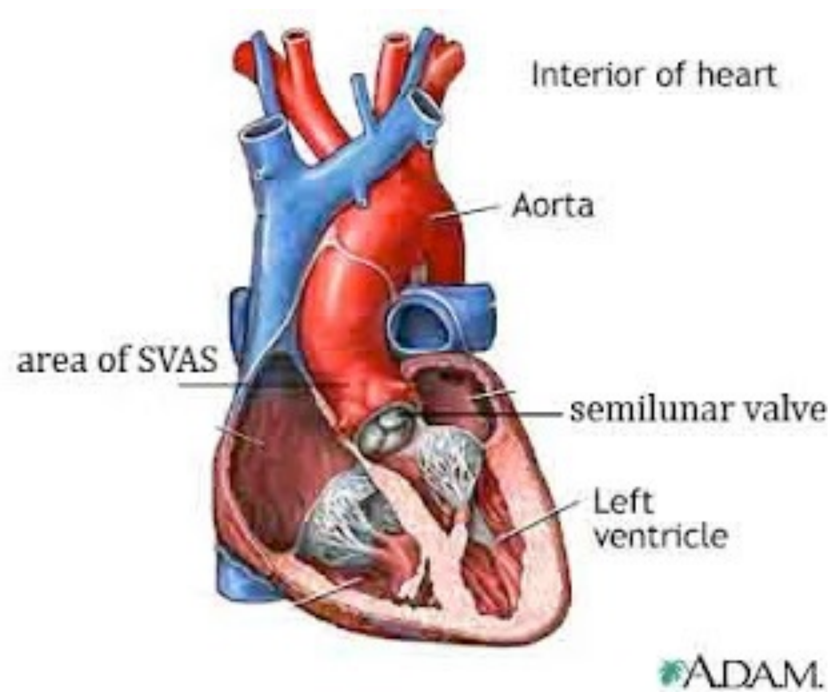
The constellation of features is often described as youthful, even for adults.

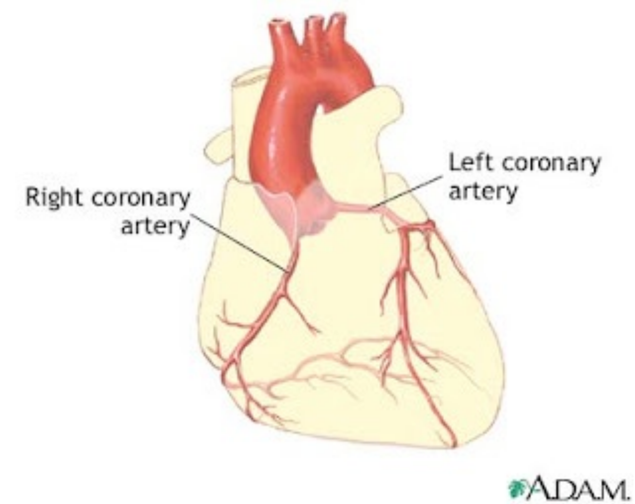


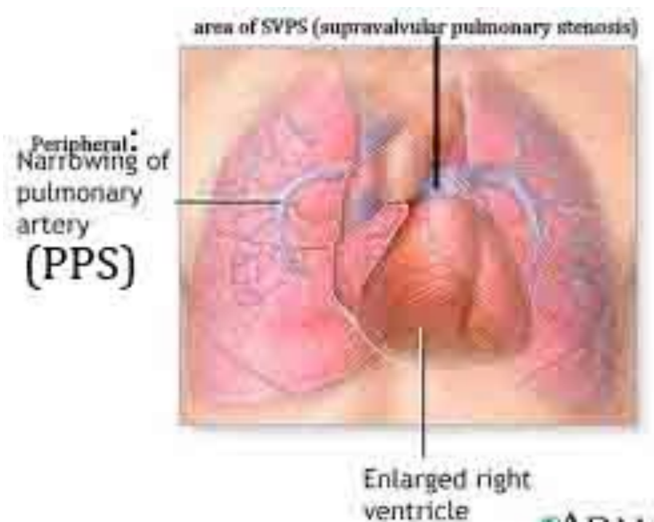
Physiological Effects: Cardiovascular

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

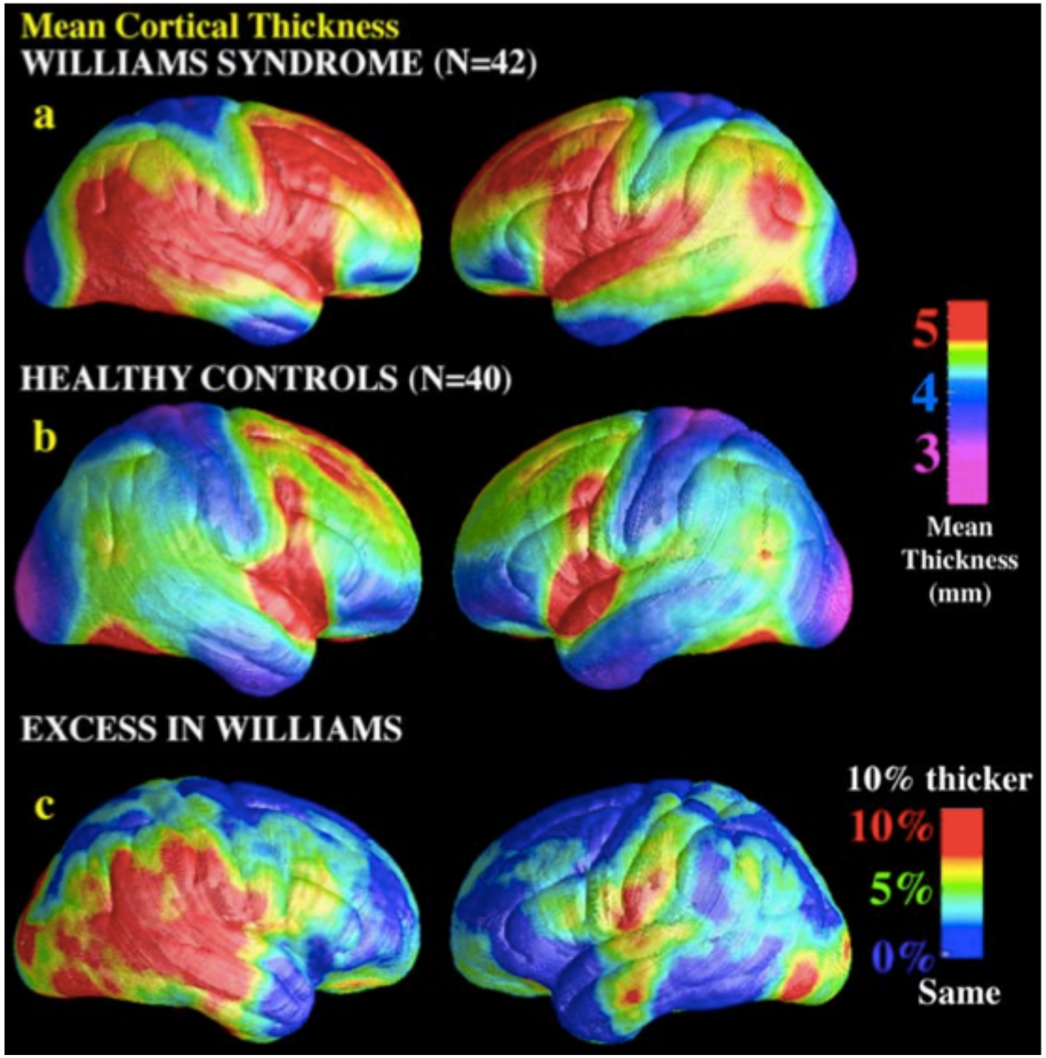


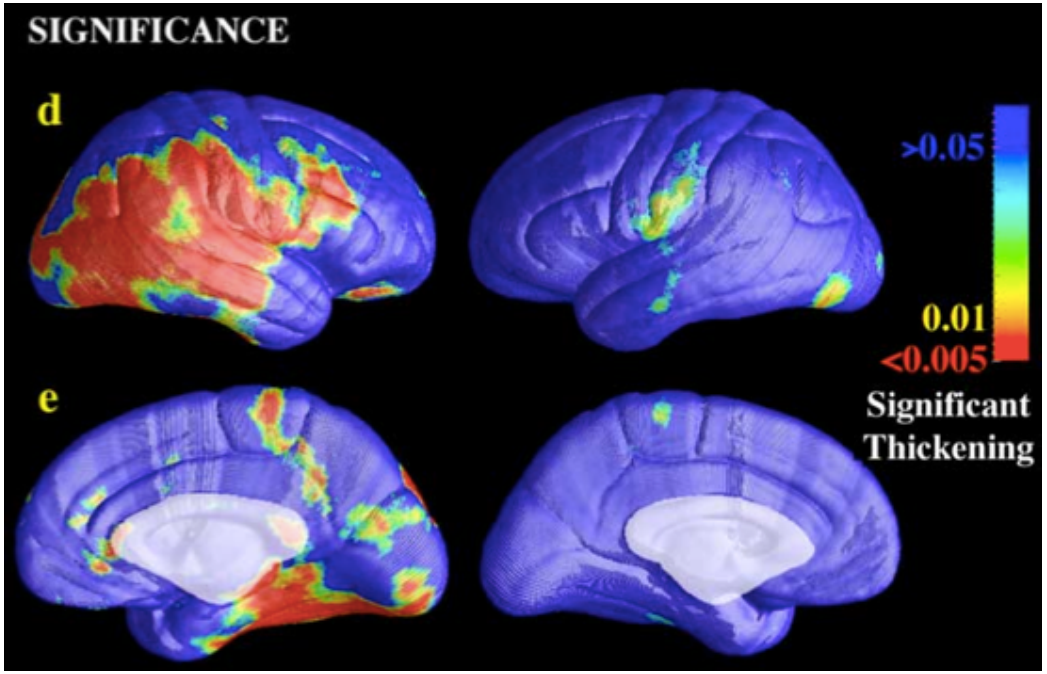




Physiological Effects: the Brain

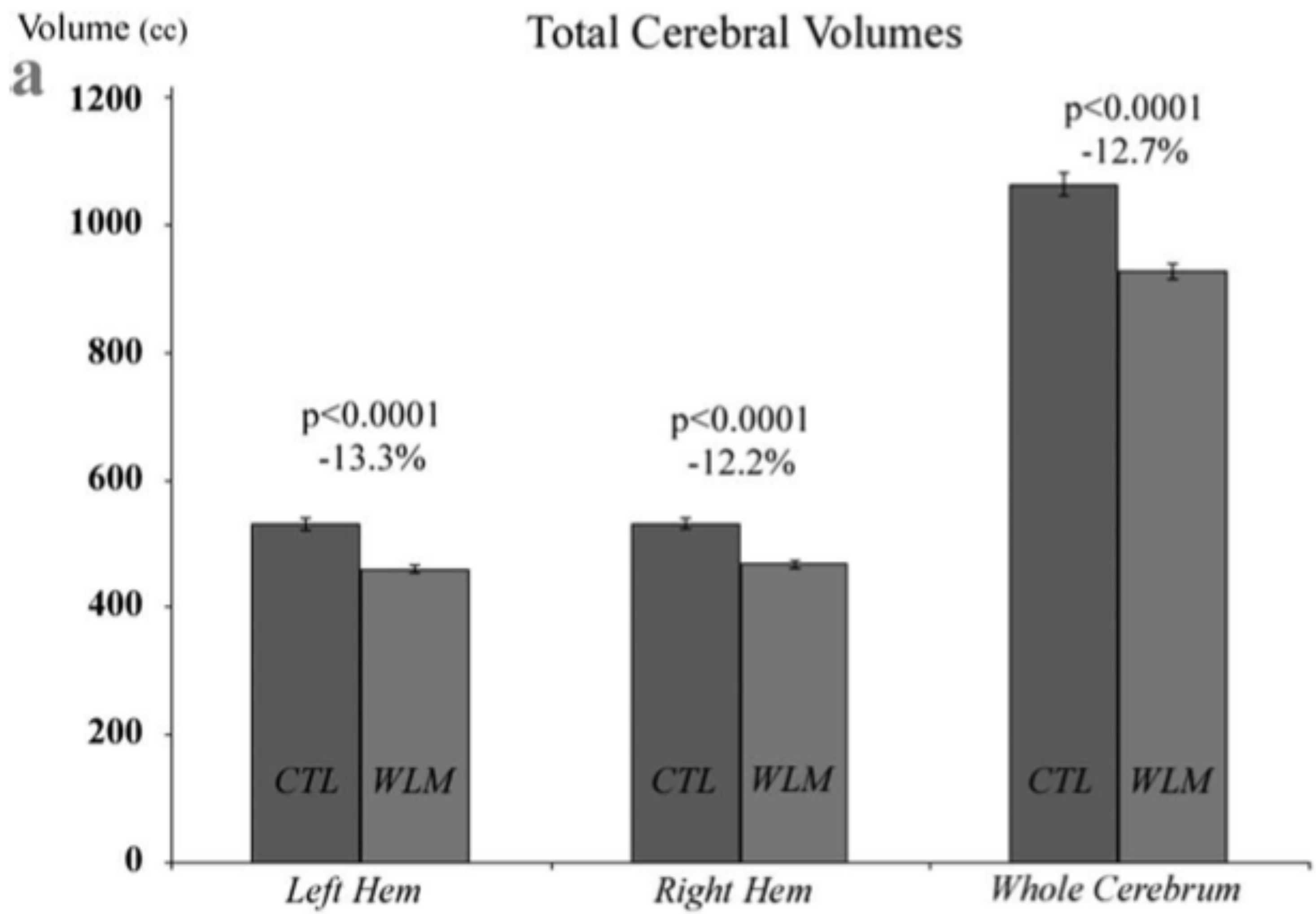
Williams Syndrome leads to a thickening of the cortex of the right hemisphere. But we have no idea how this would affect cognition.





Physiological Effects: the Brain

Williams Syndrome 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

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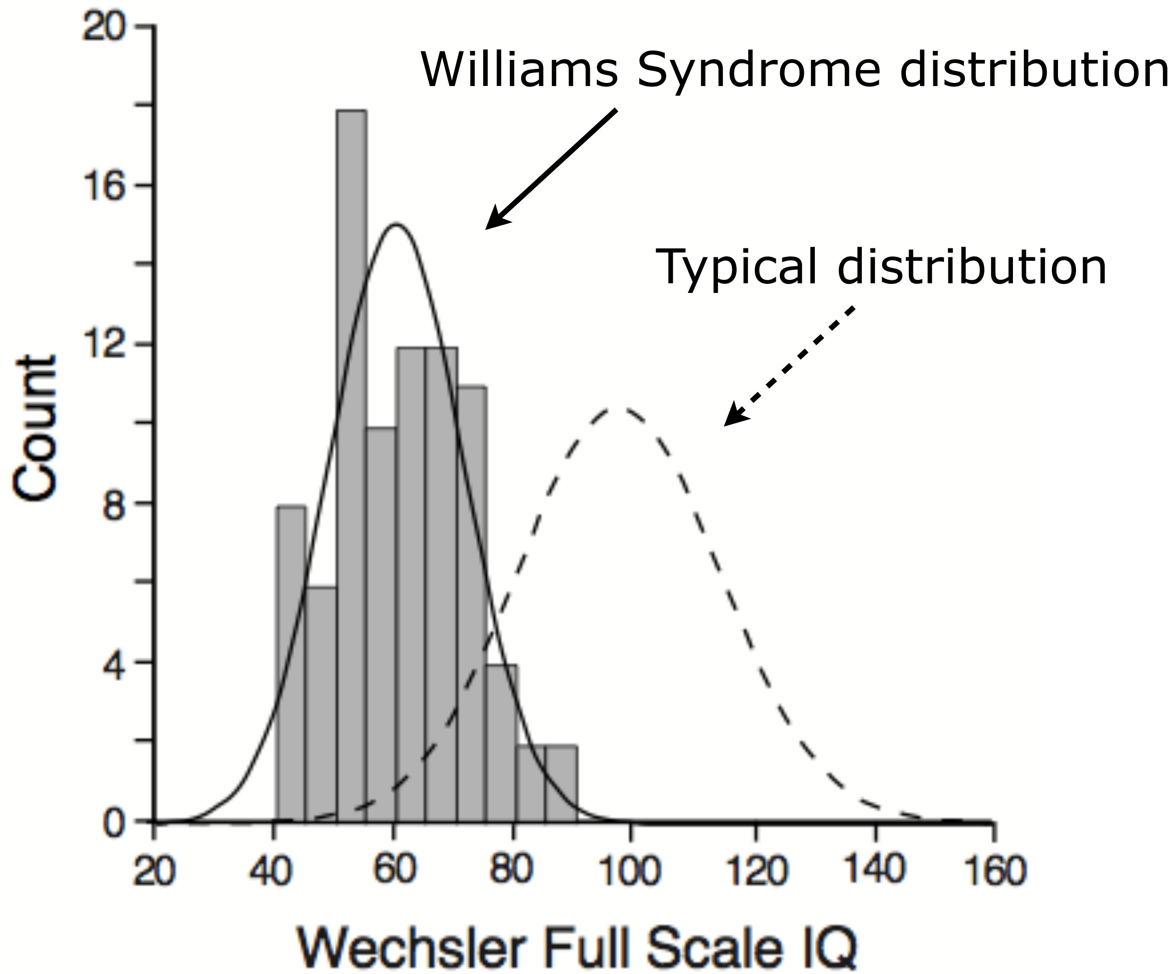
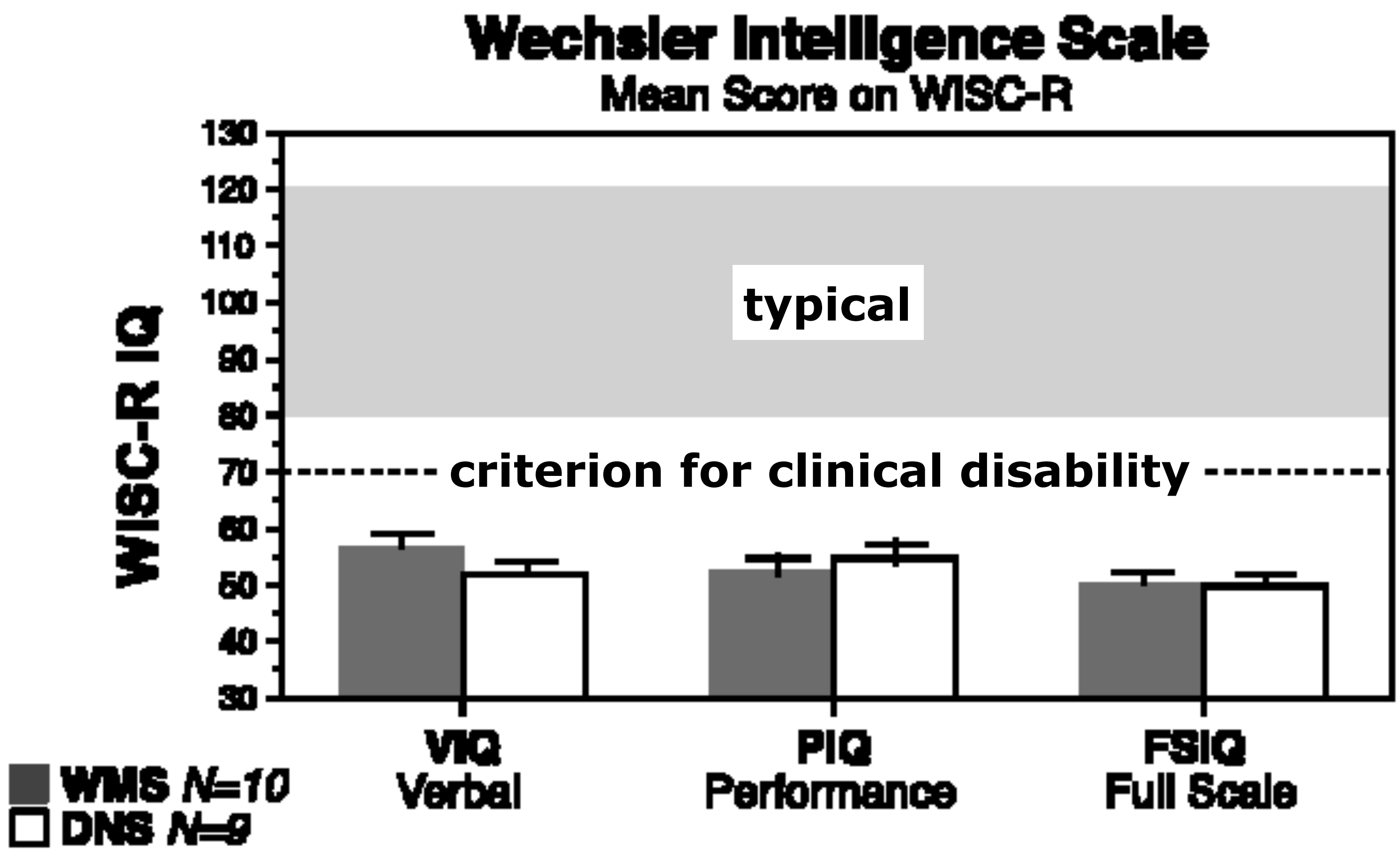


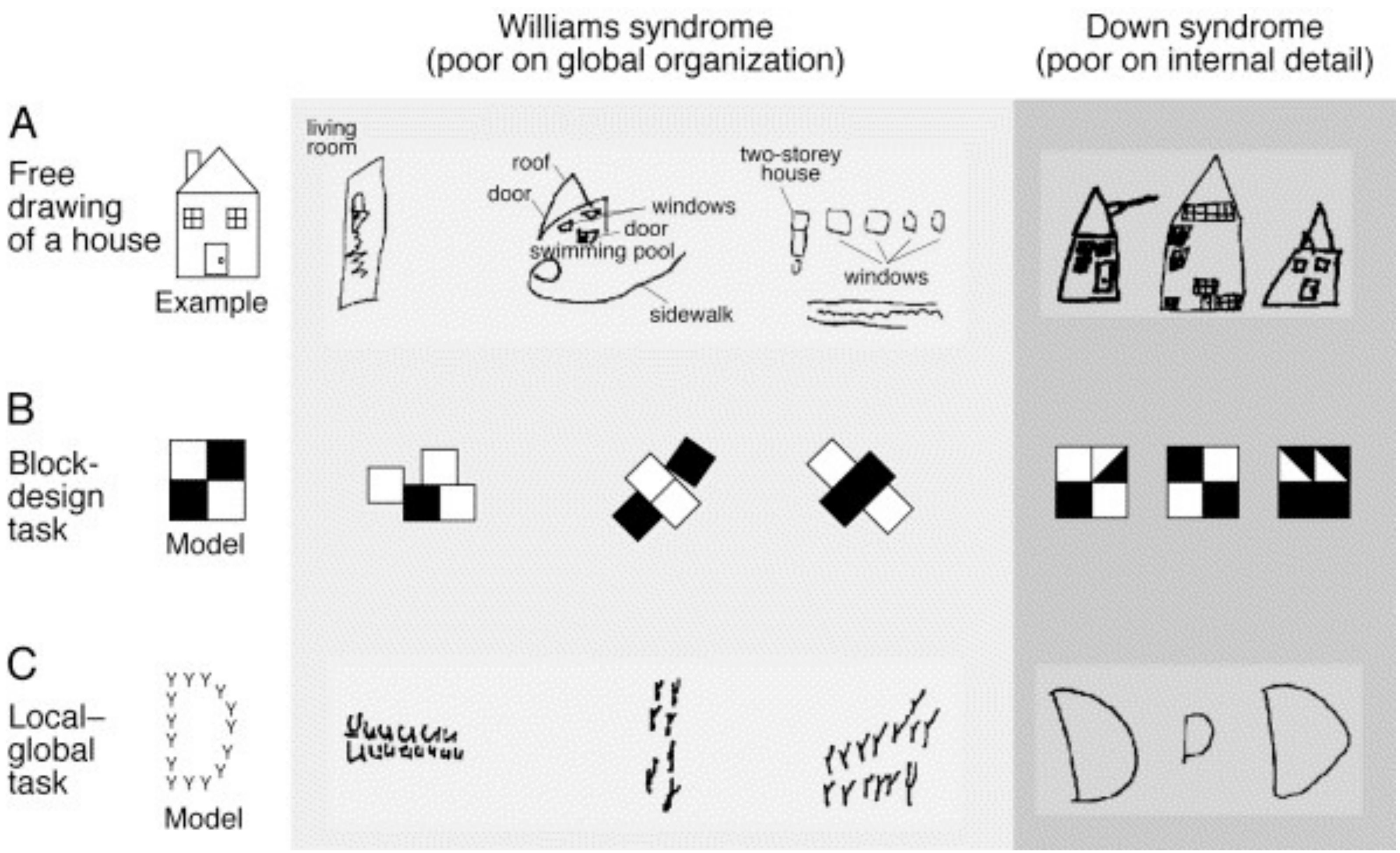
Comparing effects on intelligence in Down Syndrome and Williams Syndrome

The two disorders appear to have very similar effects on general intelligence. Keep this in mind — it means we should expect to see the same level of effect on language (if there were a link between them). But we won't!



Comparing effects on visual-spatial processing in DS and WS

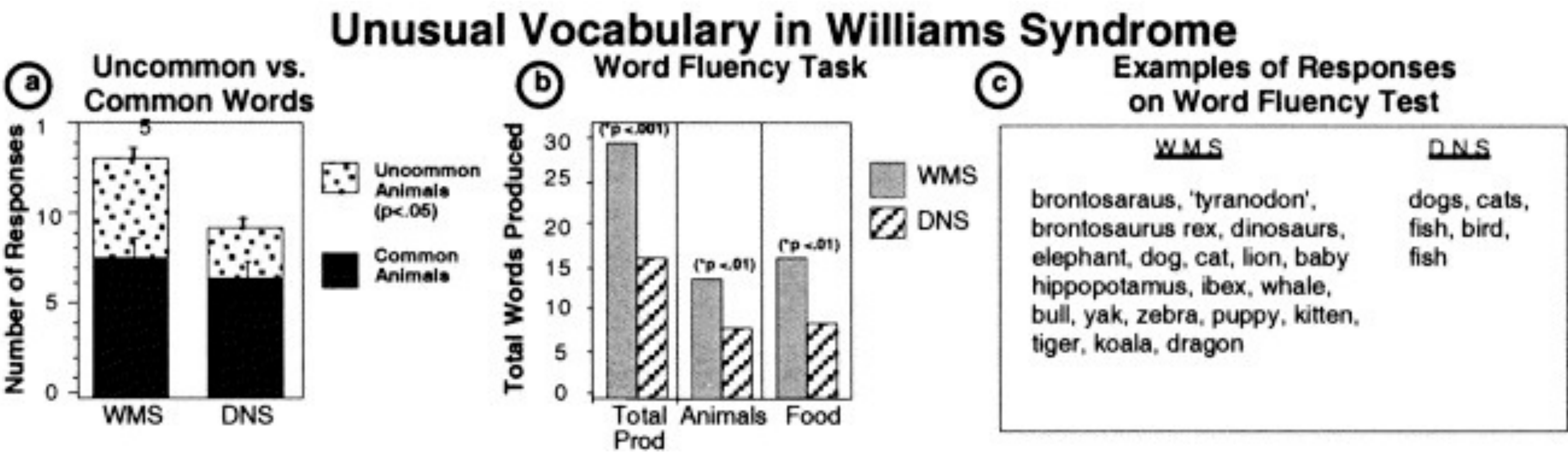
Both WS and DS lead to visual-spatial deficits. However, they are distinct deficits: WS seems to preserve internal details, but loses global organization; DS seems to lose internal details, but preserves global organization



Comparing effects on language in DS and WS

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).



Comparing effects on language in DS and WS

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

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

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."

DNS age 13

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

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 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.

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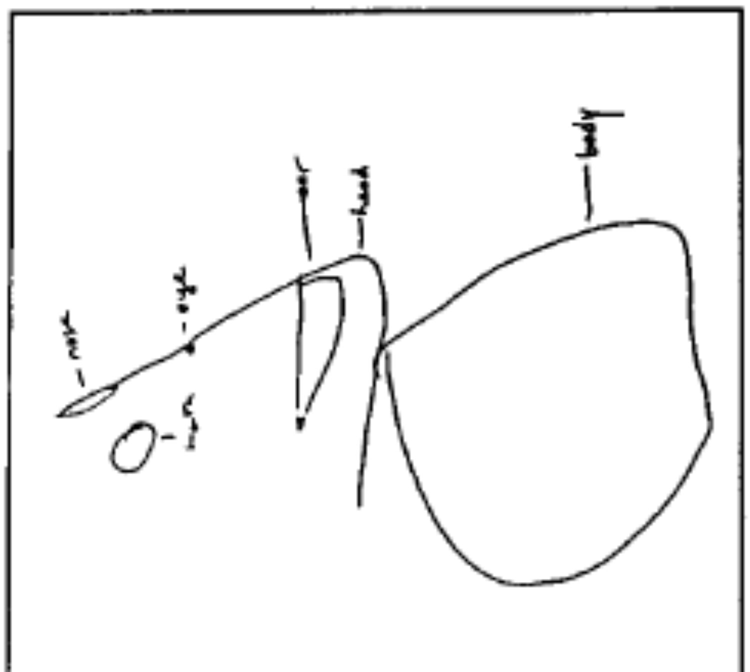
WS: comparing visual-spatial abilities and language abilities

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

a.



b.

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...

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.

This is one half of our dissociation

Williams Syndrome shows that general intelligence can be affected while language ability is (mostly) unaffected. (This also shows that the correlation seen in Down Syndrome is specific to Down Syndrome.)

Williams Syndrome	Specific Language Impairment
Affects general intelligence	Has no effects on general intelligence
Has no (or few) effects on language	Affects language acquisition

Specific Language Impairment

(or Delayed Language Disorder)

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).

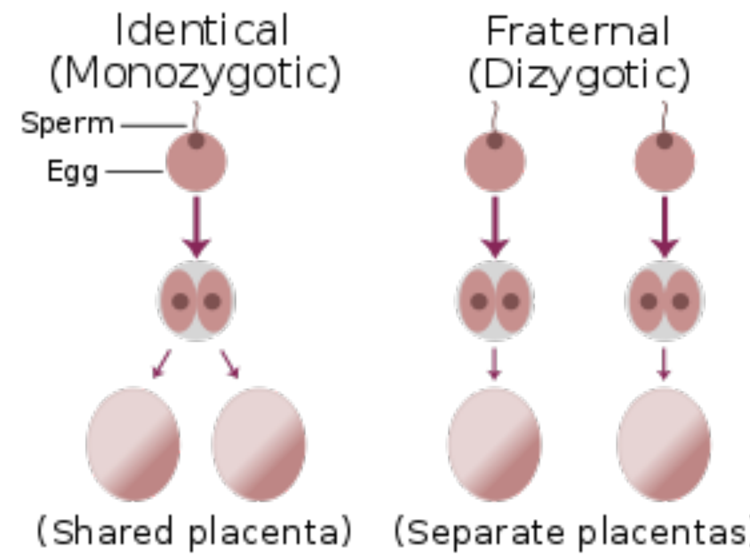
What causes SLI?

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) in which **both have SLI** is much higher than the proportion of pairs of dizygotic twins (two eggs - fraternal twins) in which both have SLI. **This suggests genes are a stronger cause than environment.**

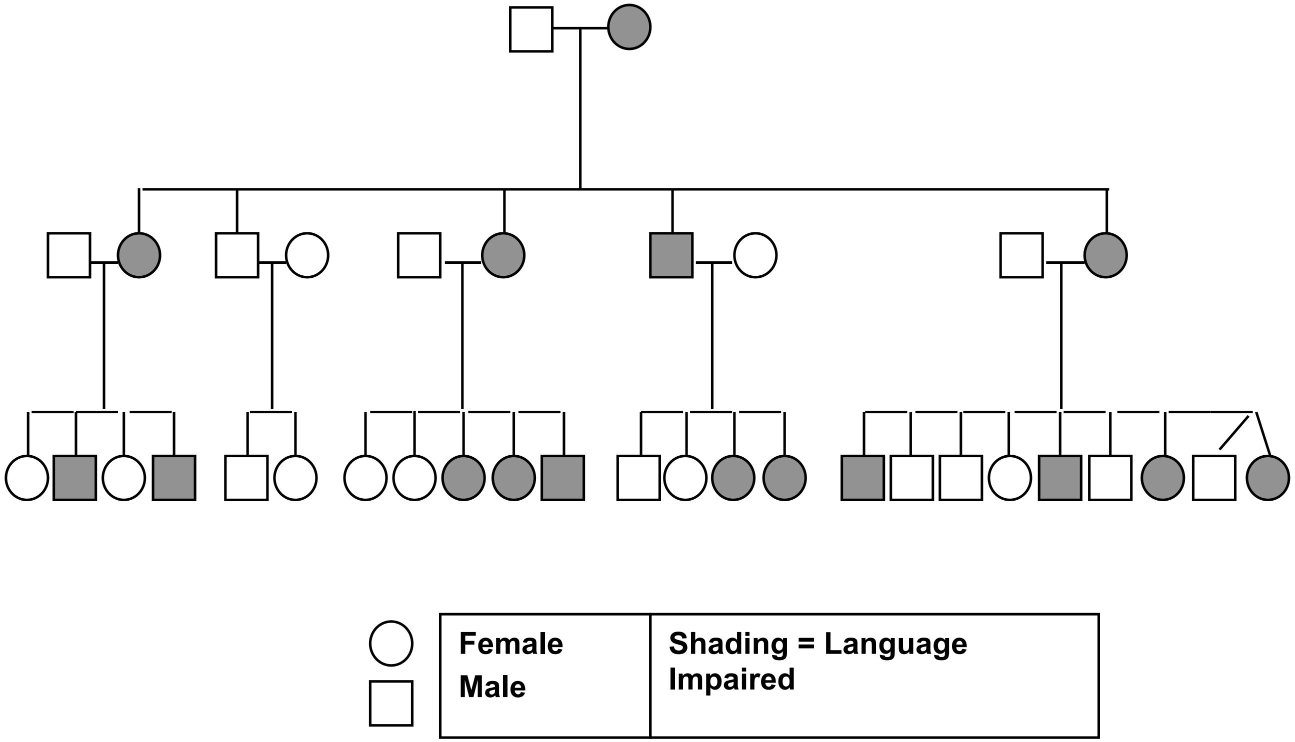
Furthermore, in cases where only one member of monozygotic twins has SLI, the other tends to show some language impairment, though perhaps not severe enough to meet the diagnostic criteria for SLI.

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:

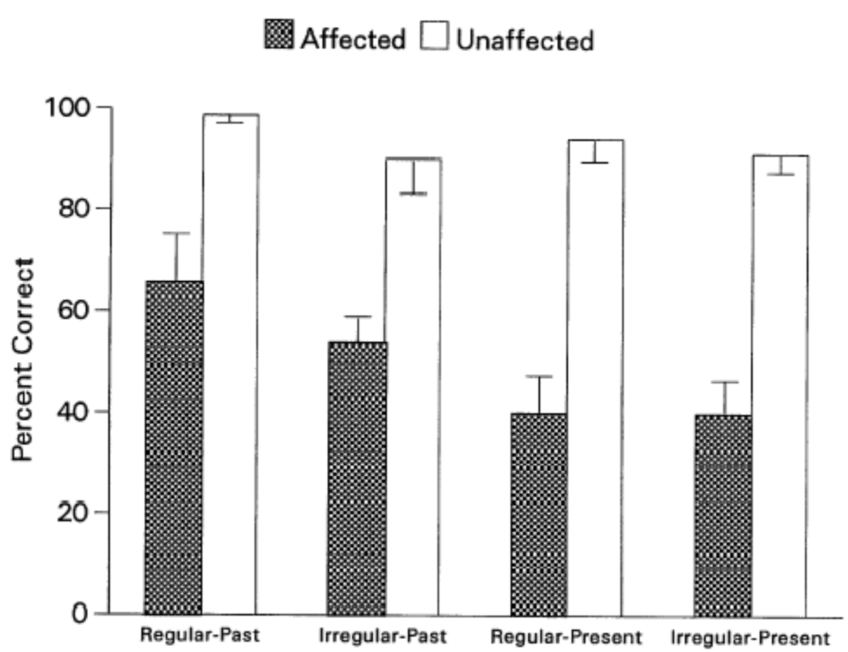


FIG. 2. Production of tenses. Scores are means ± standard errors. See Table 2 for examples of test items.

But they also show deficits in oral-facial abilities:

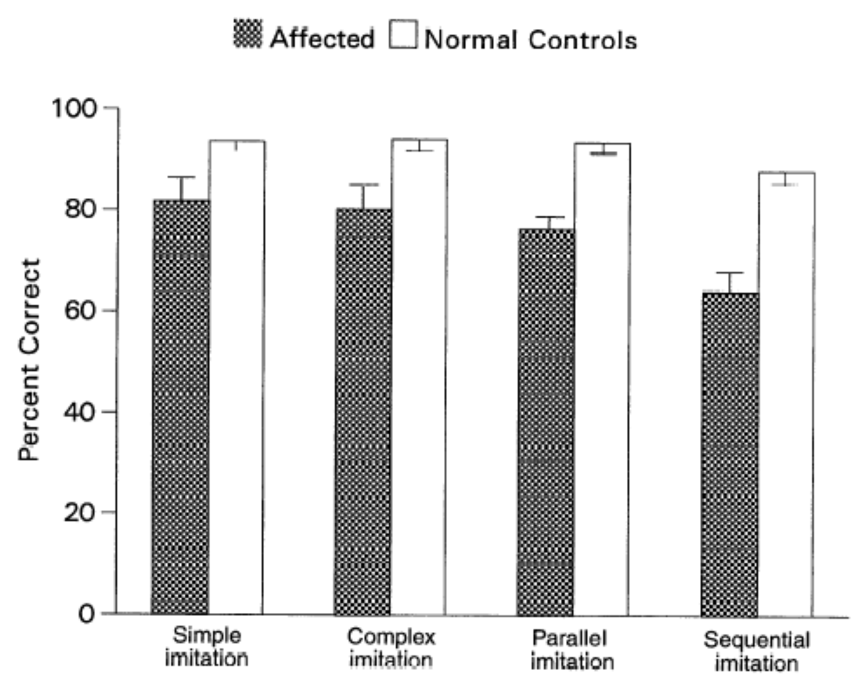


FIG. 3. Imitation of oral and facial movements. Scores are means ± standard errors.

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 of intelligence.

Williams Syndrome	**Specific Language Impairment**
Affects general intelligence	Has no effects on general intelligence
Has no (or few) effects on language	Affects language acquisition

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