MODULARITY

• Williams Syndrome (WS):
  – (more or less) general cognitive retardation except in language
  – Results from a gene deletion in chromosome 7.

• Specific Language Impairment (SLI):
  – Language problems without any obvious cause.
  – fifty to seventy percent of children with SLI have at least one other family member with the disorder

➢ If the brain is a general purpose problem solver, one’s general intelligence should be predictive of abilities such as language.

➢ For people affected by WS or SLI that’s simply not true.
Williams Syndrome

• Caused by a sporadic deletion of one copy of 20 or so genes.
• Affects about 1/20000 births, boys and girls equally.
• IQ about 50 (range 40-90) but cognitive profile very uneven.
• WS caused by the deletion of one copy of a small set of genes on chromosome 7, band 7q11.23.
• Approximately 20 genes deleted. Little variability in the breakpoints.
• For most of these genes, their role in the Williams phenotype is unknown.
• ELN: the gene for elastin, a protein that provides elasticity to organs and tissues.
• LIM-kinase 1: has been argued to be responsible for aspects of visu-spatial cognition, but this is still debated.
Williams Syndrome physical appearance

Facial Features

- full prominent lips
- stellate iris pattern
- prominent ear lobes
- wide mouth
- small, widely spaced teeth
- medial eyebrow flare
- flat nasal bridge
- short nose/anteverted nares
Williams Syndrome medical features

• Cardiovascular problems
• Developmental delay (infants height & weight < 5th percentile)
• Infantile hypercalcemia (elevated calcium level in blood)
Persons with WS are bad at:

• Spatial cognition tasks
• Coordination
• Piagetian conservation tasks
  – Ability to recognize that number, mass or volume does not alter when physical appearance alters.
  – E.g. □ = ◆

• Math and number
• Folkbiology (Johnson & Carey, 1998)
Persons with WS are bad at:

- Spatial cognition tasks
  - But they know that their models are not correct.
- Contrast between visuo-spatial and language abilities:
Persons with WS are bad at:

- Spatial cognition tasks
- Distinct modes of failure in WS and Down:
Persons with WS are good at:

• Language
• Auditory short term memory.
  – Recall: This is precisely what’s impaired in dyslexia and SLI
• Social cognition
  – overly friendly, uninhibited, personality
  – Emotional, and sensitive to emotions of others
  – Theory-of-mind
• Face processing
• Music
Persons with WS are good at:

• Music.
  – Individuals with WS generally have a higher rate of musicality.
  – WS persons tend to have an “affinity” for music -- this, however, does not necessarily entail musical talent.
  – The percentage of individuals with Williams syndrome who have perfect pitch is higher than the percentage among individuals in the general population.

• Music is often considered the most closely related cognitive ability to language.
  – E.g., ERP responses to ill-formedness in music and language similar in many cases.
Persons with WS are good at:

• Face processing.

Figure 13. The strengths and weaknesses in visuo-spatial processing in WMS show an unusual profile. The results are shown from two tasks that are both visuo-perceptual tasks, sensitive to right-hemisphere damage, where the correct answer requires only pointing to a picture without any constructional component. Note that the same subjects with WMS perform very differently on the two tasks. The contrast in performance on line orientation (Benton Judgement of Line Orientation, mean percent correct = 36.57) and face discrimination (Benton Face Recognition, mean percent correct = 92.59) is shown for 16 subjects with WMS ($t = 18.69; p < .0001$). On the Line-Orientation task, several individuals with WMS could not even pass the warm up items. In great contrast, exactly the same subjects with WMS perform remarkably well on a very difficult face discrimination task that involves recognizing the same individual under different conditions of lighting, shadow, and orientation. In both tasks, performance of normal individuals is indicated by the broken lines.
Persons with WS are good at:

• *Language*
  – Often compared to Down Syndrome as overall IQ is similar.
Individuals with Down’s and Williams Syndromes have similar IQs.
Excellent Processing of Conditionals by Individuals with Williams Syndrome

Experiment Question: "What if you were a bird?"

WMS 1: You could fly, you could have babies, fly north or south, east or west.

DNS 1: Bird seeds.

WMS 2: Good question. I'd fly through the air being free.

DNS 2: You'd be strong.

WMS 3: I would fly through the air and soar like an airplane and dive through trees like a bird.

DNS 3: I don't fly.

WMS 4: I would fly where my parents could never find me. Birds want to be independent.

DNS 4: I not a bird, you have wing.

WMS 5: I would fly and if I liked a boy, I would land on his head and start chirping.

DNS 5: Fly in the air.
WS Language

• However, whether language is really spared has been heavily debated.

• For example, it’s been reported that WS kids perform badly on embedded relative clauses. (Karmiloff-Smith et al, 1997; Volterra et al., 1996; Mervis et al., 1999)
  – but who doesn’t?
    • [The juice [that the child spilled ]] stained the rug.
What does the test look like?

“The circle the star is in is red”
• Hardly surprising if a person with general mental retardation does not do so well on this task.
• Challenge: to devise tasks that tap onto language without being taxing for other cognitive resources.
Example
(from Andrea Zukowski’s work)

Method: Elicited production technique
(Hamburger and Crain, 1982)

Children are asked to tell a parent which of two similar characters a change happened to.
Which cow is Max looking at?
“The cow who um the boy’s pointing to.”

(12-year-old Williams Syndrome male, IQ = 56)
(12-year-old Williams Syndrome male, IQ = 56)

"The cow who um the boy’s pointing to."
Which boy turned blue and which boy turned purple?
“The boy that’s pointing to his arm turned purple, and the boy that turned blue is pointing to his finger”
Subject
Gap
Relatives

“The boy that’s pointing to his arm turned purple, and the boy that turned blue is pointing to his finger”
“The boy that’s pointing to his arm turned purple, and the boy that turned blue is pointing to his finger”
What was each WS individual capable of producing?

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<th>CE Relative</th>
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## Control Children

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Every WS child except one was capable of producing examples of each of the components of embedded relative clauses.
WS and language

• WS kids produce complex syntactic structures as long as the task demands are otherwise low.

• At the lexical level, WS individuals tend to show an absence of typical frequency effects.
Anomalous Semantic Processing in Williams Syndrome: Homonyms Task

(a) Stimulus Examples

E: Which two go together best?
- BANK (Target)
- MONEY (Primary Associate)
- RIVER (Secondary Associate)

(b) Performance on Similarity Judgment Task

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(c) Definitions of Homonyms

E: What does "nuts" mean?
- DNS: "We crack nuts." (Probe) "We eat nuts."
- WMS: "There are two kinds of nuts, a peanut, and nuts and bolts."

E: What does "club" mean?
- DNS: "Go to a club." (Probe) "I'm in the key club."
- WMS: "A secret kind of club, and a club with spurs-those pointy things for killing animals."

Figure 6. (a) The figure shows stimulus examples of homonyms task with primary and secondary meanings (e.g., 'bank'). (b) Subjects with DNS and normal mental age controls provide more primary meanings than secondary meanings of the homonyms, as would be expected. Subjects with WMS, in contrast, provide an equal number of primary and secondary meanings, suggesting anomalous semantic organization. (c) Sample responses show that WMS subjects are able to access both the primary and secondary meanings of homonyms while DNS subjects access only one meaning, sometimes with two responses. For the meaning of 'club,' a DNS individual said "Go to a club" and then "I'm in the key club" whereas a WMS individual said: "A secret kind of club, and a club with spurs ..."
WS brains

- Total volume of the brain is reduced.

WS brains

- Somewhat conflicting results on regional preservation and enlargement.
- Some studies have reported preserved or even enlarged frontal areas, neocerebellum, temporal limbic areas and superior temporal areas but a large-scale 2005 study did not replicate most of these findings.
Neocerebellum

- Involved in movement.
- Among the brain's newest parts, appearing in human ancestors about the same time as the enlargement of the frontal cortex, which controls much of rational thought and reasoning.
- The neocerebellum is significantly smaller in people with autism, who are generally antisocial and poor at language, the reverse of people with Williams.

Limbic system

- memory, emotions
Robust finding: thicker RH cortex in WS

- Classically right-lateral functions that WSs are **good** at:
  - music
- Classically right-lateral functions that WSs are **bad** at:
  - Spatial cognition

- Studies such as this one constitute important progress in solving the mystery of WS but they do not point to any simple explanation to understanding the complex behavioral profile of WS.
• No significant thinning.