

# Developmental Disorders affecting language

Williams Syndrome

# Williams Syndrome (WS)

- Discovered in 1964
  - Infantile hypercalcemia (excess calcium)
  - 1/7500 children
- Genetic Disorder
  - Deletion/mutation of genes on chromosome 7q11.23
  - Many protein deficiencies
  - Elastin is a protein found throughout the body



Figure 1 Fluorescent in situ hybridization (FISH) of the ELN cosmid (arrow) and a chromosome 7-specific cosmid at D75396 (7q36) with metaphase chromosomes of an individual with WS, demonstrating deletion of ELN on one homologue (line) but not the other (arrowhead).

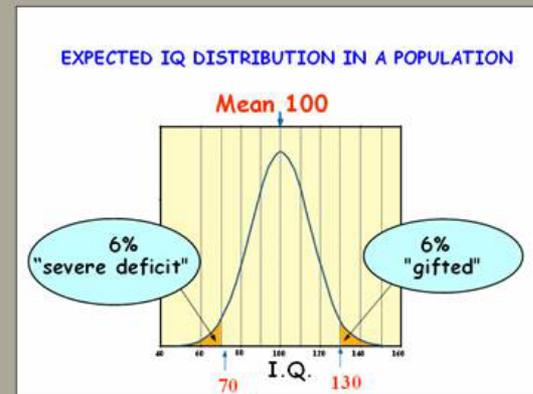
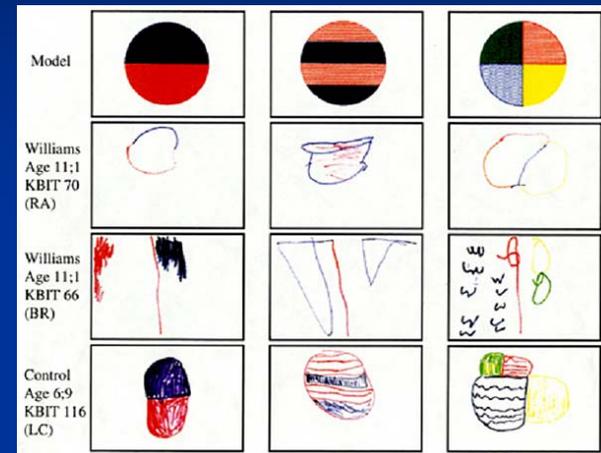
# Medical Phenotype

- Characteristic facial appearance
- Abnormal growth pattern
  - Low birth weight
  - Early puberty
  - Shorter than normal growth spurt in puberty
- Hypercalcemia (excess calcium)
- Hypotonia; hyperreflexia
  - Cerebellar problems
- Ophthalmologic problems
- Hypersensitivity to sound
- Dental problems
- Connective tissue abnormalities
  - Skin, joints
  - Cardiovascular
- Gastrointestinal problems
- Urinary tract abnormalities
- Musculoskeletal problems



# Cognitive Phenotype

- Relatively good auditory rote memory
- Relatively good language
  - **But** delayed acquisition!
- Very poor visuo-spatial abilities
- Highly social/overfriendly
  - “delightful”
- Attention deficit disorder
- Anxiety
- Full-scale IQ around 50
- IQ excluding spatial tests (K-BIT) ranges from 40-102; mean 66



# K-BIT (non-spatial) IQ

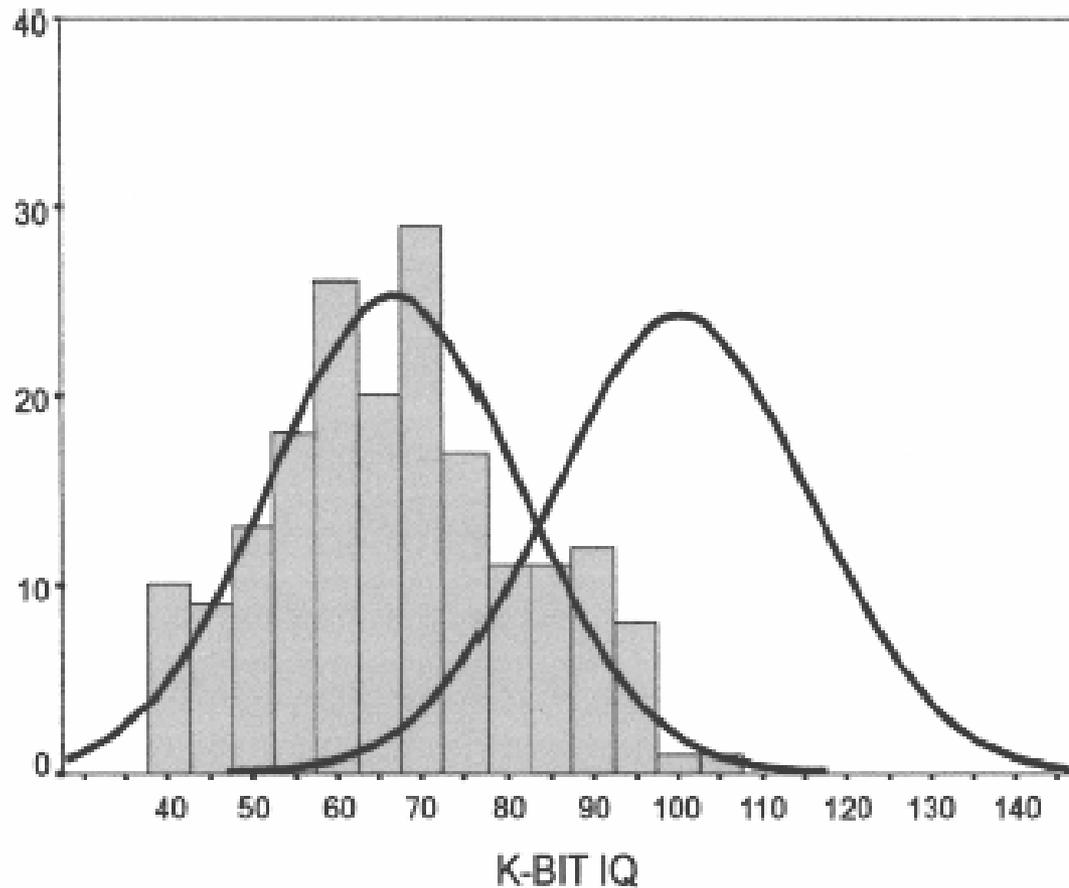


Figure 3 Bell-shaped distribution of K-BIT IQ scores for a sample of 186 individuals with WS (on the left) compared to the bell-shaped IQ curve for the general population (on the right). For the sample with WS, mean IQ is 66.12, whereas mean IQ for the general population on this test is 100. The variability in IQ evidenced among individuals with WS occurs even though 98% of individuals with WS have the same size deletion.

# Visuo-spatial abilities

- Very poor
- Focus on details
- Poor on block design
- Not a deficit of visual perception
  - Perception not worse than age and IQ-matched controls
  - Face perception seems to be relatively strong

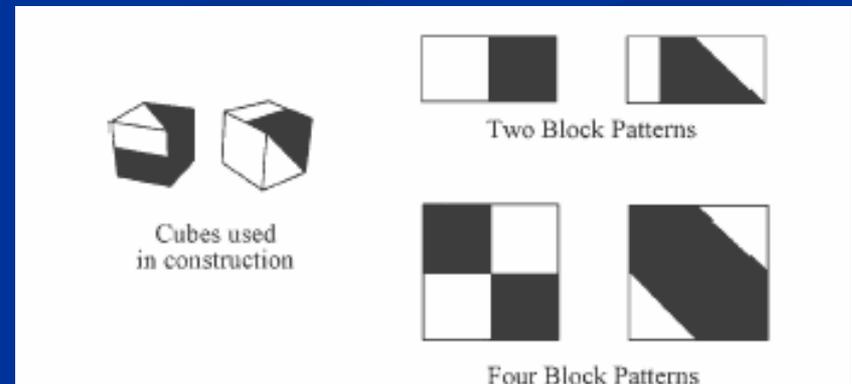
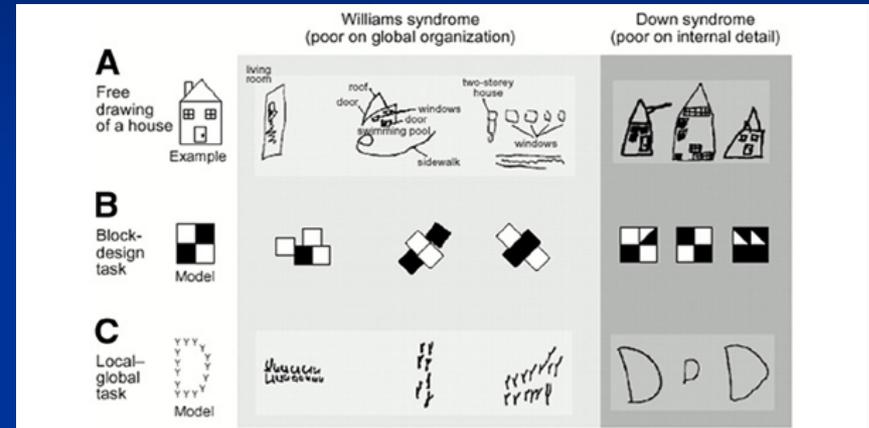


Figure 4 Cubes used for the DAS Pattern Construction subtest, and examples of easy (left) and relatively difficult (right) patterns that could be constructed with these cubes. Each cube has one solid yellow side (depicted as white in the figure), one solid black side, two sides divided diagonally into yellow and white triangles, and two sides divided vertically into yellow and black rectangles. Note that the participant is always given the correct number of cubes for the pattern he or she is to construct.

# Brain Findings

- Overall reduction in volume (but small size in general, not just brain)
- Controlling for overall volume
  - Reduced volume
    - Parietal cortex
    - Corpus callosum
    - Brainstem
  - Increased volume
    - Auditory cortex (temporal lobe)
    - Cerebellum
- Increased cortical gyrification/complexity
- Abnormal hippocampal shape
- Atypical neuron size and packing

# 2 Theories (1)

- Language Modularity
  - Preserved language in Williams Syndrome suggests that language is independent of cognition
- What is a module?
  - Specialized, encapsulated processor
- Granularity problem
  - What is modular about language?
    - All of it?
    - Just Syntax? Morphology? Phonology?
    - Particular morphosyntactic rules?

# 2 Theories (2)

## ■ Neuro-constructivist

- Williams syndrome is a developmental disorder
- Language in Williams syndrome reflects altered constraints on cognitive and linguistic development
- Early language studies important, before compensation can take place

## ■ Two hypotheses

- Over-reliance on spared phonology
  - (phonological short term memory)
  - Lexical-semantic processing is impaired
- “Conservative hypothesis”
  - Language is delayed but normal

# A difficulty...

- What is benchmark for 'normal' language?
  - Chronological-age matched typical controls?
    - But large IQ differences
  - Mental-age matched typical controls?
    - But large chronological age differences
  - Chronological and mental-age matched atypical controls?
    - Other disorders may be different in many ways

# Language Profile in Williams Syndrome

Is it really spared?

# Pragmatics

- Use of language in a social context
- Early studies suggest spared pragmatics
- BUT
  - Old fashioned/formal style
  - Failure to respond adequately to questions
  - Focus on irrelevant topics in conversation
  - WS make fewer inferences about mental states than controls
  - “cocktail party speech”
    - Fluent speech; overfamiliar manner, etc.
  - Impaired at figurative/non-literal language
    - Lies, jokes

# Syntax

## ■ Receptive

- Normal comprehension of
  - Reflexives ('himself')
  - Passives
  - Complex embedded clauses
- Receptive syntax worse than receptive vocabulary

## ■ Expressive

- Parent interview indicates comparable complexity of sentences in WS and typical controls, both better than Down syndrome
  - Spontaneous speech samples suggests normal complexity and mean length of utterance (MLU)
  - Sentence repetition impaired in Italian
  - Impaired elicited production of relative clauses
- ## ■ Apparently no compensation by rote learning

# Morphology

- Regular vs irregular plural or past tense
  - English, German, Hungarian
- Relative to matched control subjects
  - Regulars seem to be largely spared
    - Possibly impaired in some cases
  - Irregulars seem to be impaired
  - Novels seem to be spared in some cases, impaired in others

# Phonology

- Phonology in WS
  - Speech perception biased towards prosody
    - Trouble (delays) with syllables/phonemes
  - Speech production appears normal
    - Normal relationship with motor development
- Phonological short term memory
  - “peak ability”?
  - Better at digit span and word span than Down syndrome (weakness)
  - Not better than typical controls

# Phonological Fluency/Awareness

- FAS Fluency
  - Performance comparable to age/IQ matched controls
- Rhyme detection (identify word pairs that rhyme)
  - Williams subjects worse than controls matched on reading/vocabulary
- Phoneme Deletion (delete first sound in word)
  - Williams subjects worse than controls matched on reading/vocabulary

# Vocabulary Acquisition

- Delayed acquisition of first words in WS
  - Commensurate with global cognitive delay
- Difficulty with joint attention
  - Important for vocabulary learning
  - Considered part of pragmatic deficit in autism
- Older WS children and adults perform better on receptive vocabulary test than expected based on mental age
  - Children with Down syndrome were worse than expected
- Not better than controls on productive vocabulary measures (e.g., naming)

# Other lexical processes

- Homonym processing
  - Bank = money / river
  - Given three words, which two go together
    - WS showed reduced bias towards frequent meaning
      - Deficit?
      - WS more able than controls to produce infrequent meaning when asked
- Semantic priming
  - Appears to be normal in WS

# Summary of Language Profile

- Pragmatics
  - Tentatively impaired
  - May be similar to autism
- Grammar
  - Commensurate with overall cognitive ability
  - Possibly impaired; possibly not atypically developing
- Phonology
  - Relatively spared
  - Difficulty segmenting speech stream
- Lexicon
  - Strong receptive
  - Atypical/weak expressive?

# Brainman Movie

# Questions to Consider

- Do you see these characteristics in Daniel?
  - All savants have exceptional memory
  - All have obsessive-compulsive tendencies
  - Some savants have talents in multiple areas
- Which theories of savant talent might or might not explain his abilities? (if any)
  - Excessive practice
  - Exceptional rote memorization
  - Creation of a rich knowledge base through implicit learning
  - Compensation
  - Stronger than normal episodic memory
  - Weak central coherence