

# Cognitive and motor development in infants and toddlers with Williams Syndrome

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## Prof. Annette Karmiloff-Smith (1938-2016)



# Cross-syndrome comparisons



Dr. Dean D'Souza

Prof. Annette Karmiloff-Smith



## Down syndrome (DS)

1 in 700  
Trisomy 21  
Low IQ, speech delay



## Fragile X syndrome (FXS)

1 in 4000 males, 1 in 8000 females  
FMR1 gene mutation → FMR1 protein  
Attentional deficits, autistic traits, low IQ, speech delay

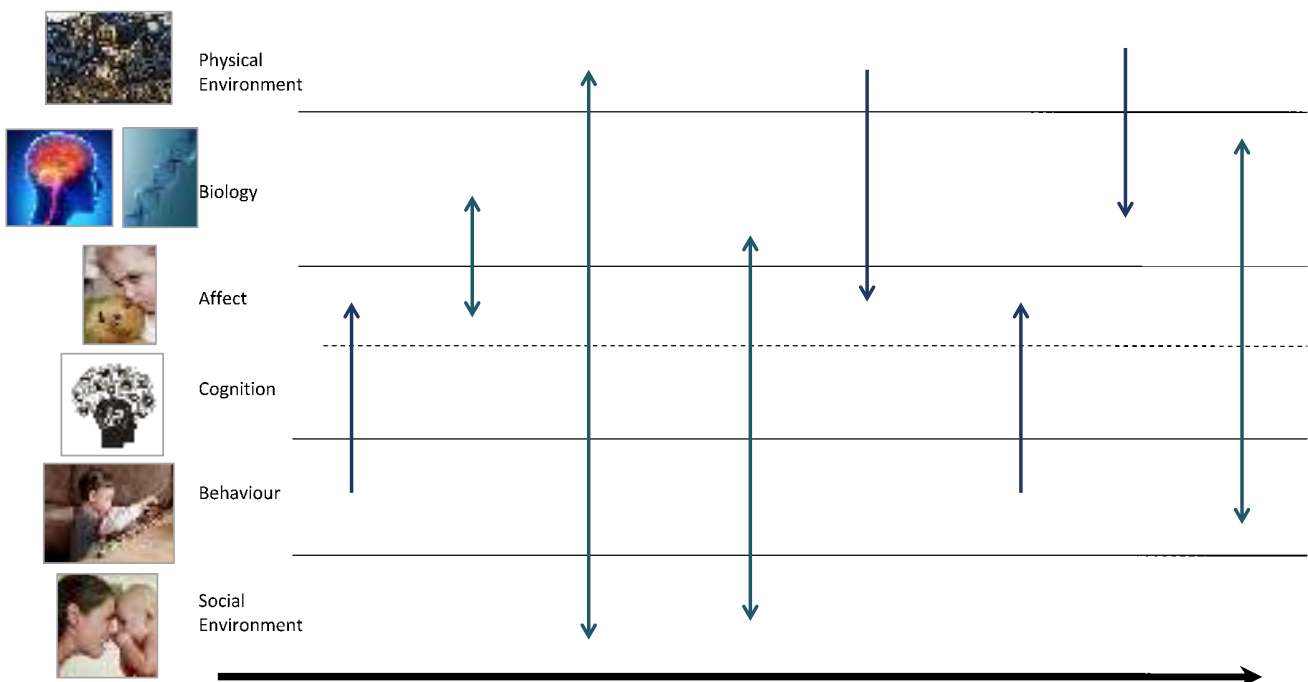


## Williams syndrome (WS)

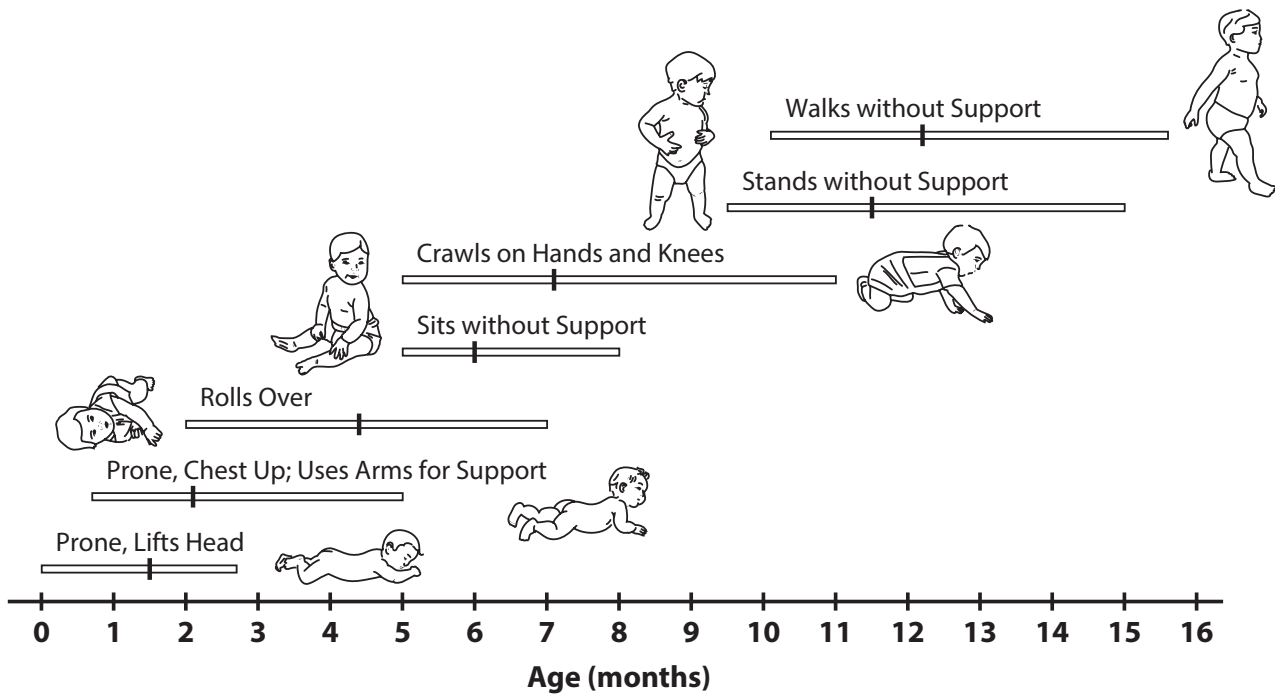
1 in 7,500-20,000  
Deletion ~26 genes, 7q11.23  
Hyper social, poor visuospatial, low IQ, speech delay

# ACORNS

Accessible Cause-Outcome Representation and Notation System

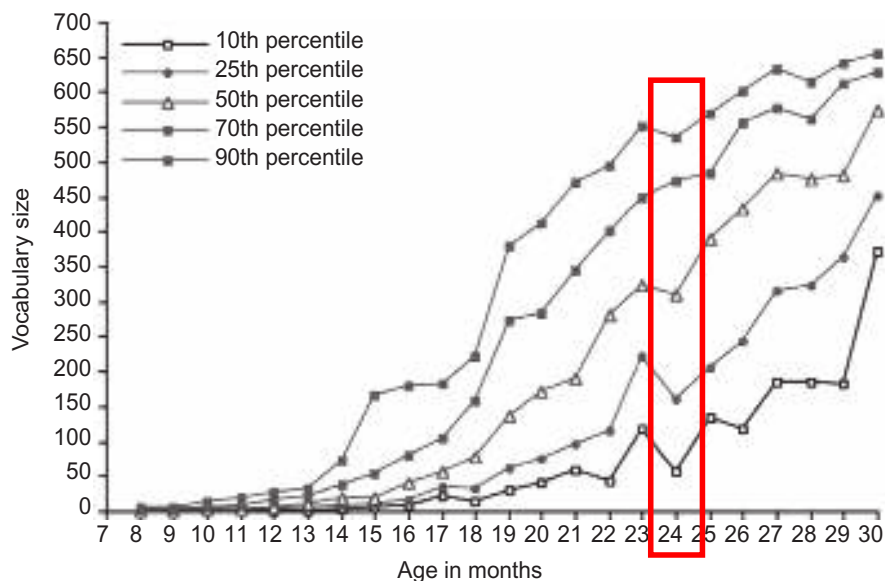


# Individual differences in typical development



Adolph & Robinson, 2015

# Individual differences in typical development



Fenson et al., 1994

# Understanding atypical development

Group comparisons:

- Typical vs. atypical group
- Cross-syndrome comparisons

Vast individual diff. in behaviourally defined disorders:

- Autism spectrum disorder (ASD)
- Attention-deficit/hyperactivity disorder (ADHD)

What about genetically defined disorders?

- **Williams syndrome?**

## Williams syndrome: typical descriptions in literature



### Genetically:

deletion of ~26 genes on chromosome 7

### Physically:

heart problems  
distinctive facial features

### Neurally:

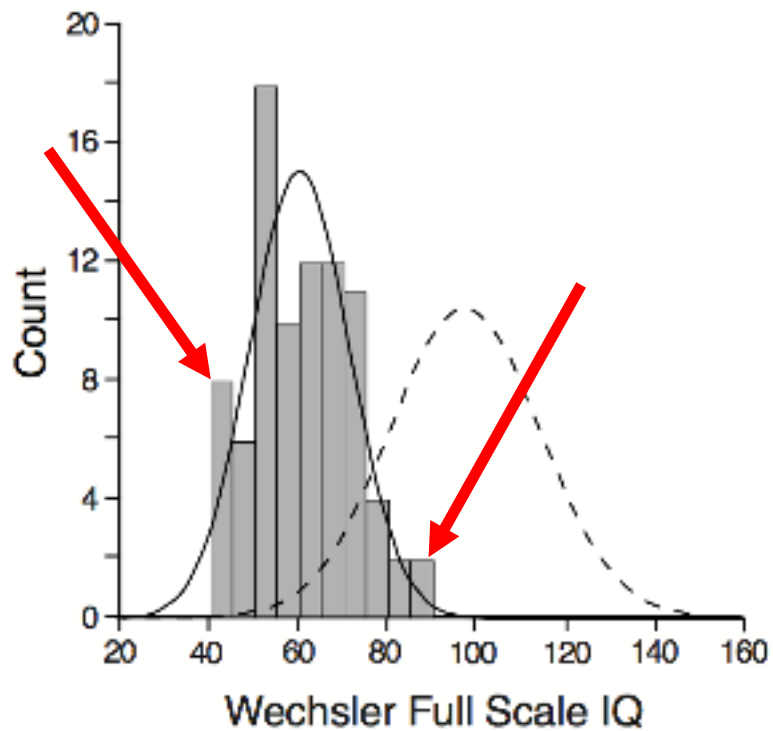
white matter reduction across lobes  
reduced gray matter in superior parietal lobe areas  
less brain activity in intraparietal sulcus

### Cognitively:

mild to moderate intellectual disability  
visuospatial processing < mental age  
language > visuospatial/number ability

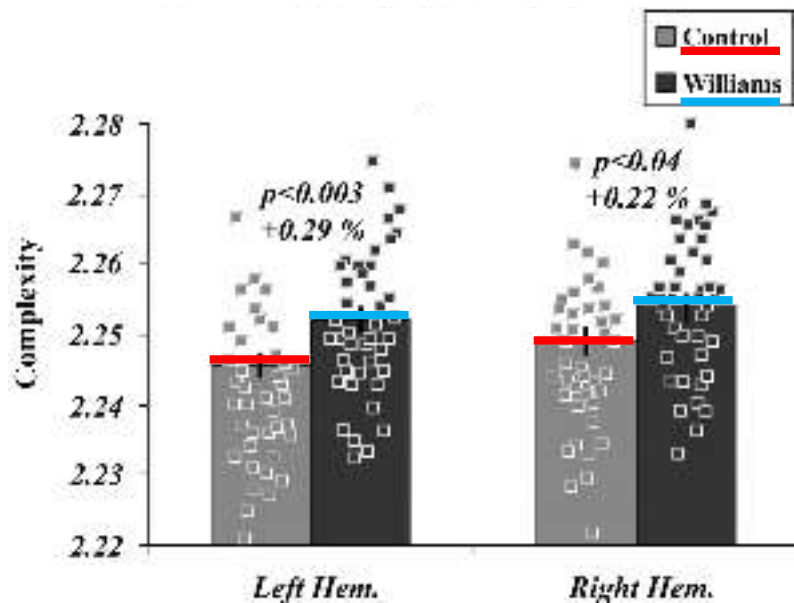
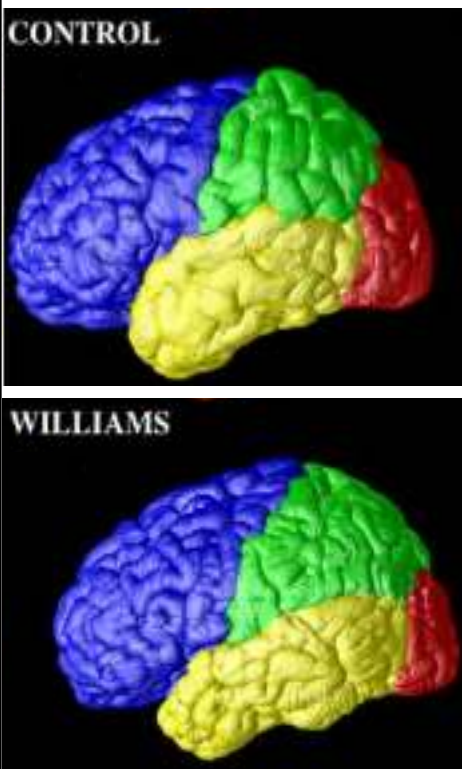
**Does this hold for *all* individuals with WS?**

# Individual differences in Williams syndrome



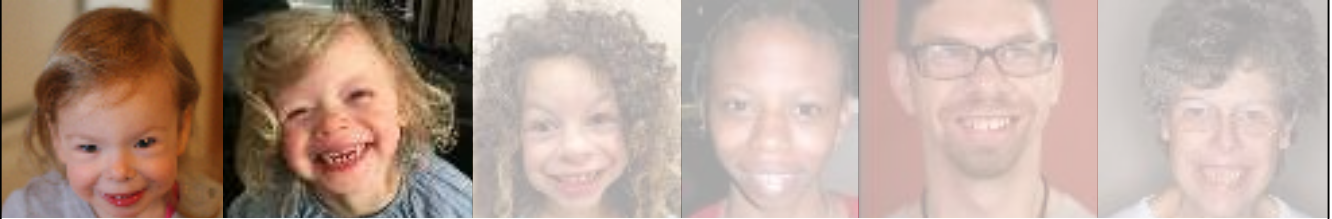
Bellugi et al., 1999

# Individual differences in Williams syndrome



Thompson et al., 2005

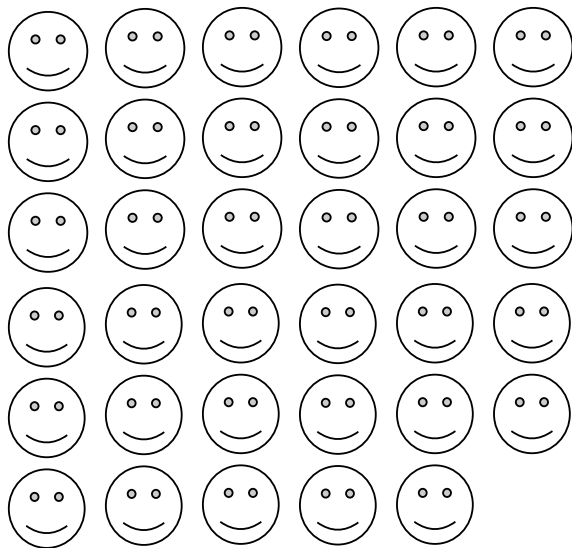
# Our aims



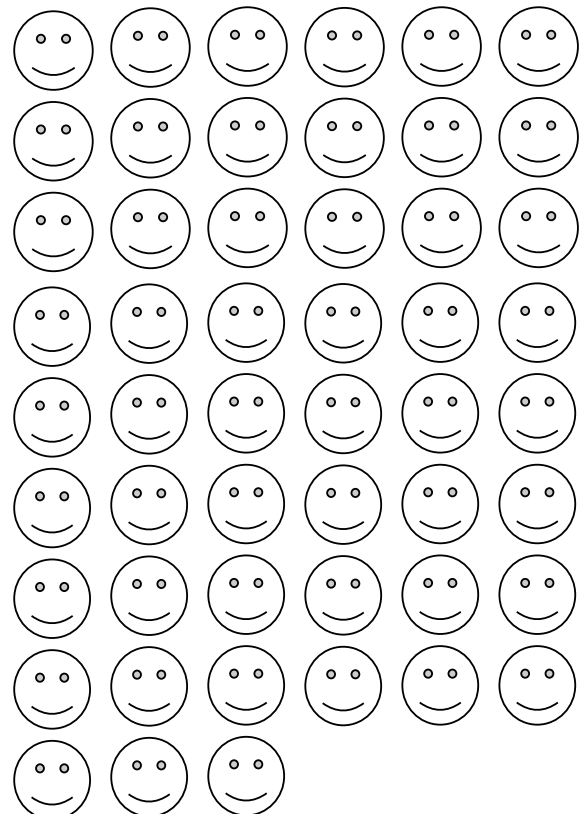
Early development

Individual differences across development

**35 WS**



**51 TD**



# Centre for Brain & Cognitive Development



## A lot to do...

**At home**



**At the visit**

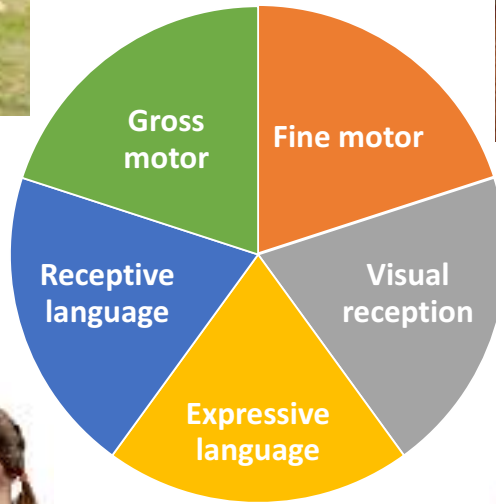


## Mullen Scales of Early Learning

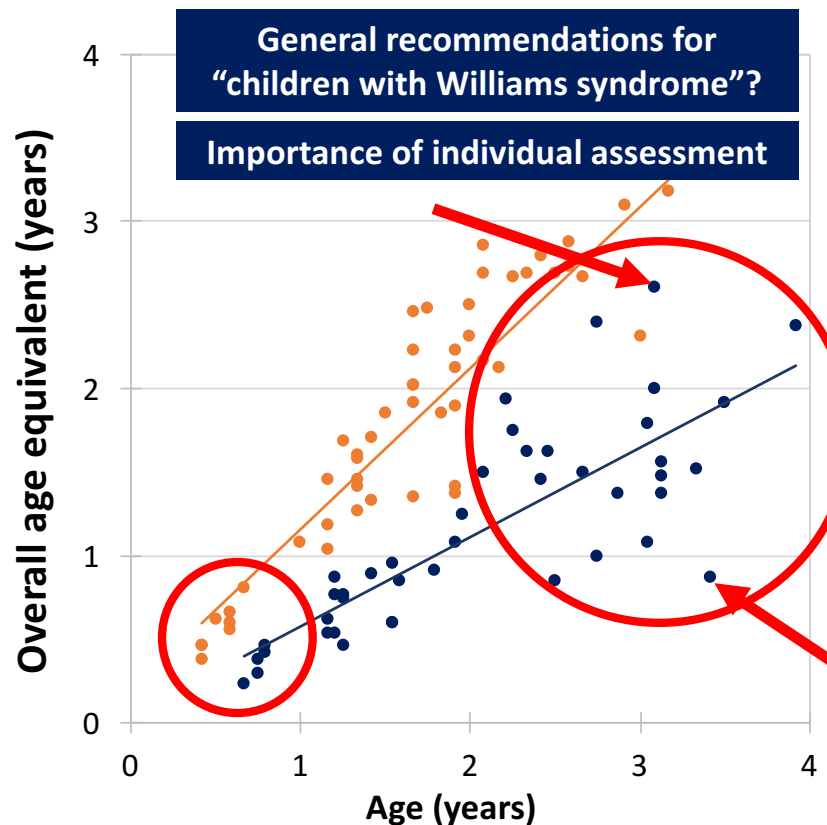




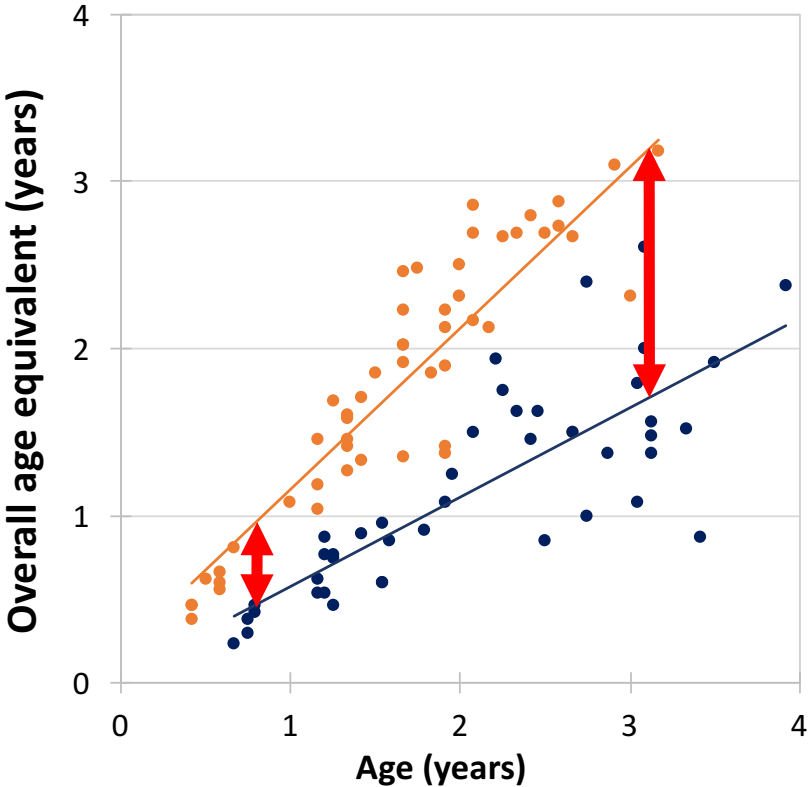
# Mullen Scales of Early Learning



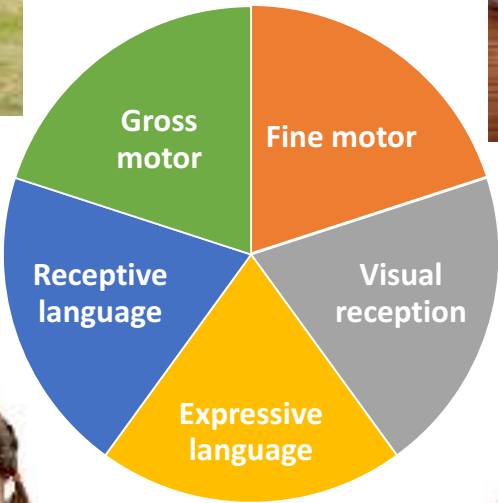
# Overall age equivalent



# Overall age equivalent



# Mullen Scales of Early Learning



# Mullen Scales of Early Learning

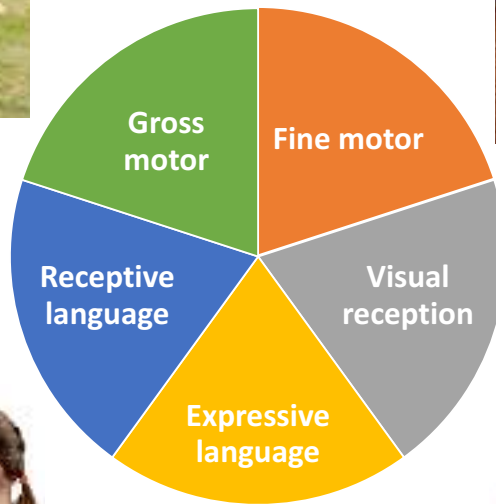
Fine motor



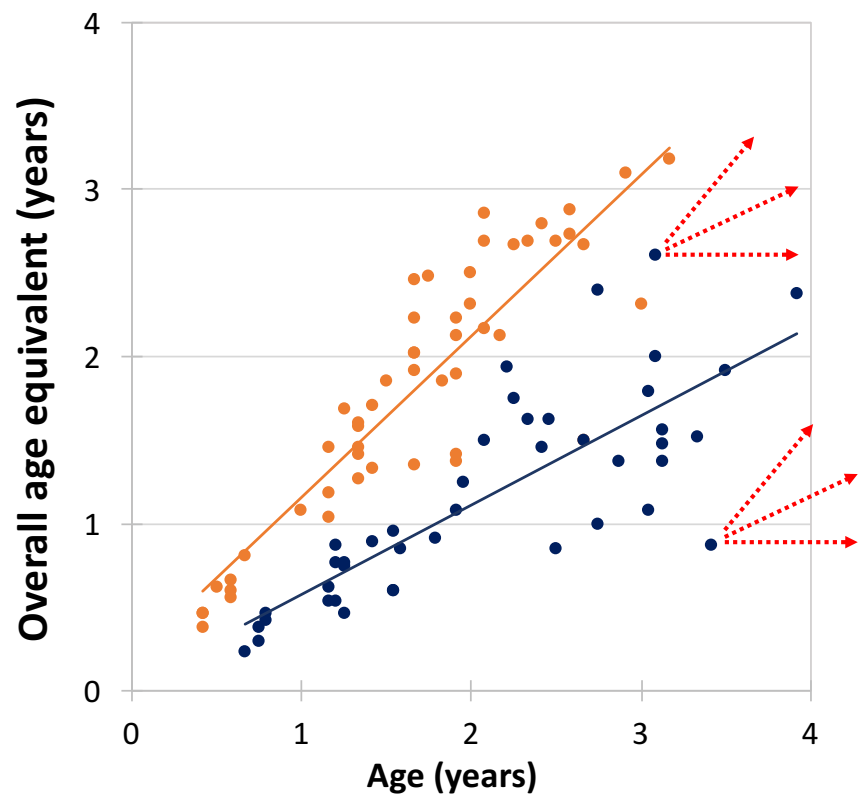
## Fine motor abilities



## Next steps...



## Next steps...



## Next steps...



Anglia Ruskin  
University

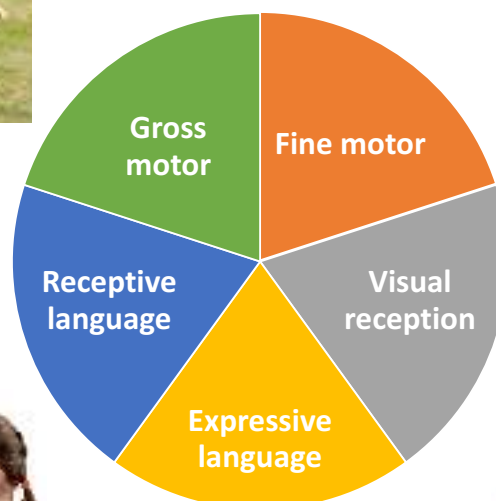


### EARLY PREDICTORS OF LATER DEVELOPMENTAL OUTCOMES IN WILLIAMS SYNDROME

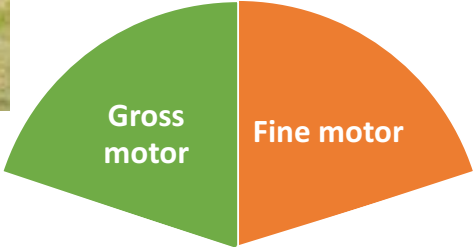
Our goal is to trace the developmental trajectories of later emerging, higher-level abilities (e.g., language) back to their basic-level roots (in, e.g., early visual attention) in children with Williams syndrome. This is a longitudinal study (from 8 months to 9 years of age).



## Mullen Scales of Early Learning



# Mullen Scales of Early Learning



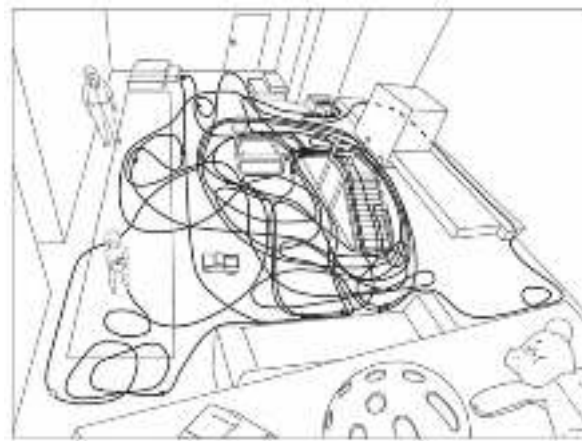
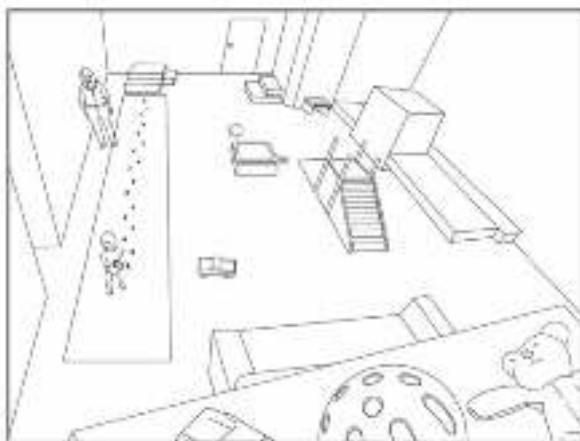
# Motor development at *core* of development itself



## The couch-potato infant



## Motor development in TD children

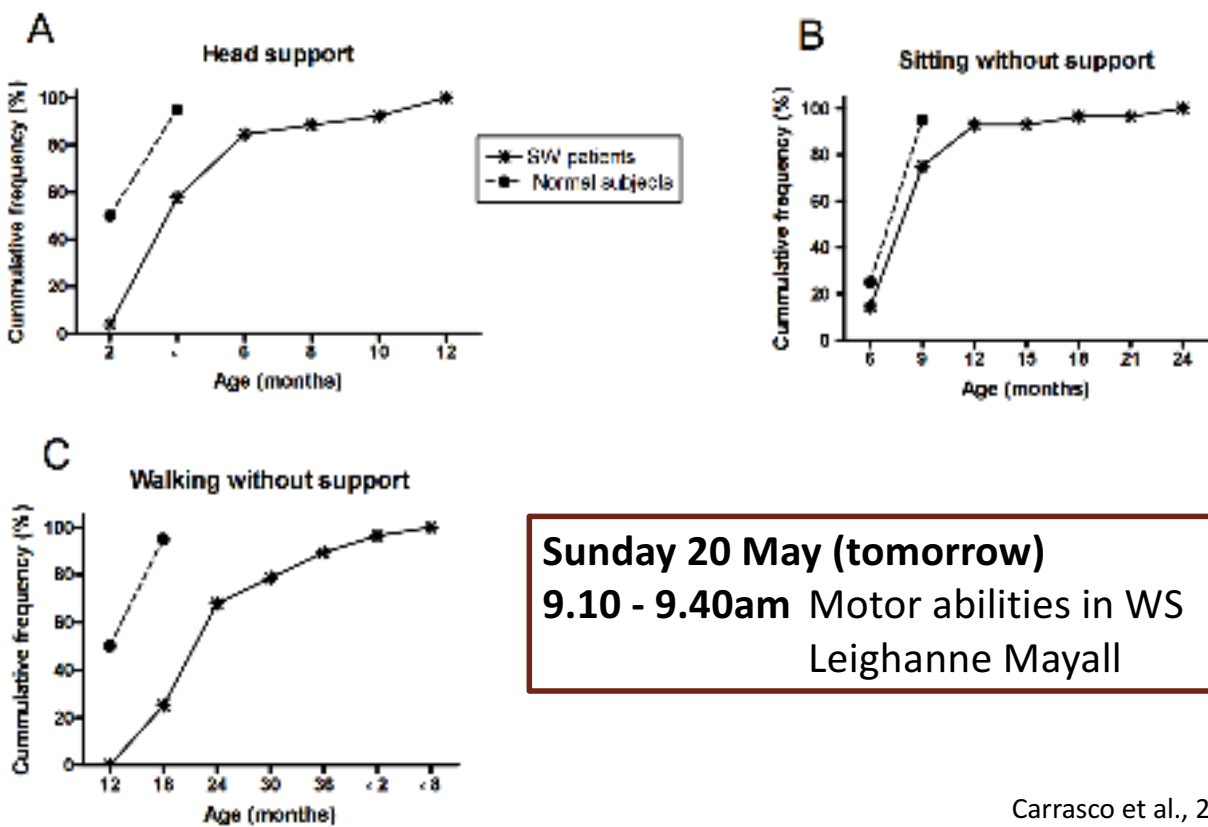


12- to 19-month-olds per hour

- averaged **2,368 steps**
- **17 falls**

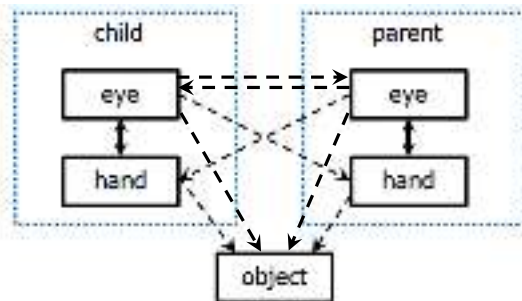


# Motor development in Williams syndrome



# Learning in the real world

Typically developing child





## The child's problem



Gavagai!

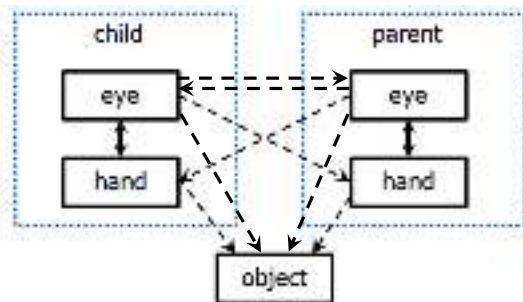
## Stepping into children's shoes...



Pereira, Smith, & Yu, 2014  
Samuelson & McMurray, 2016

# Learning in the real world

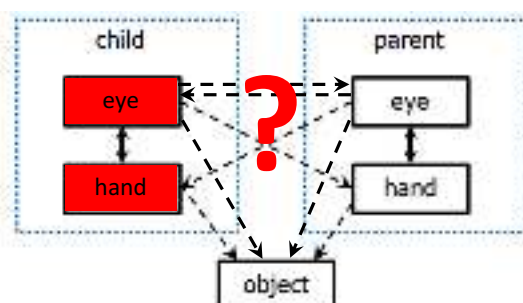
Typically developing child



Yu & Smith, 2013

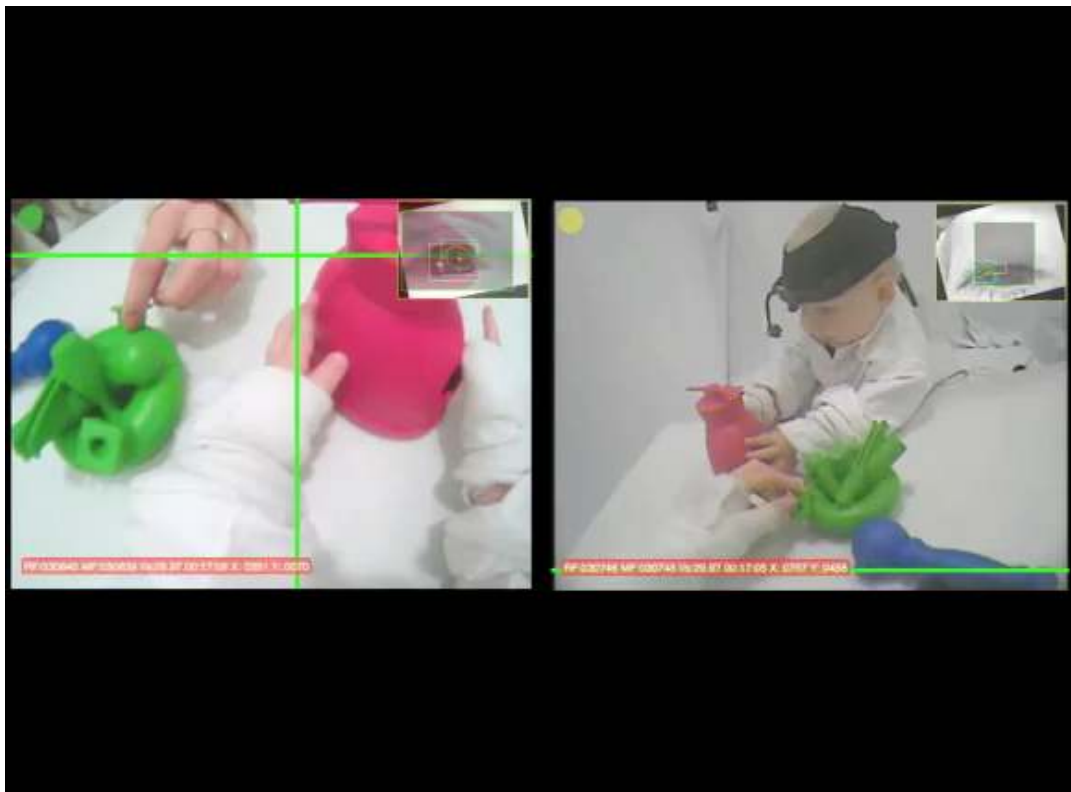
# Learning in the real world

Child with attention and motor difficulties



BETTER DATA

LABORATORY OF COGNITIVE AND PERCEPTUAL SCIENCE



## The world is a busy place full of things to learn about...

Would you like to help us discover how young children with Williams syndrome learn best?

Do you have a child with Williams syndrome under 5?

For more information, please contact Dr. Hana D'Souza:  
[hana.dsouza@gmail.com](mailto:hana.dsouza@gmail.com)

# Thank you to all our participants



BETTER DATA

THE BRITISH ACADEMY FOR THE HUMANITIES AND SOCIAL SCIENCES



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Thomas



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