The importance of cross-syndrome studies for understanding Williams syndrome

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Plan:

-Four examples of cross-syndrome studies:

Number

Attention

Face processing

Language

-The importance of *cross-syndrome, cross-domain, cross-developmental* studies

Cross-syndrome studies of number

Williams/Down syndromes

WS/VCFS(22q)

WS/FXS

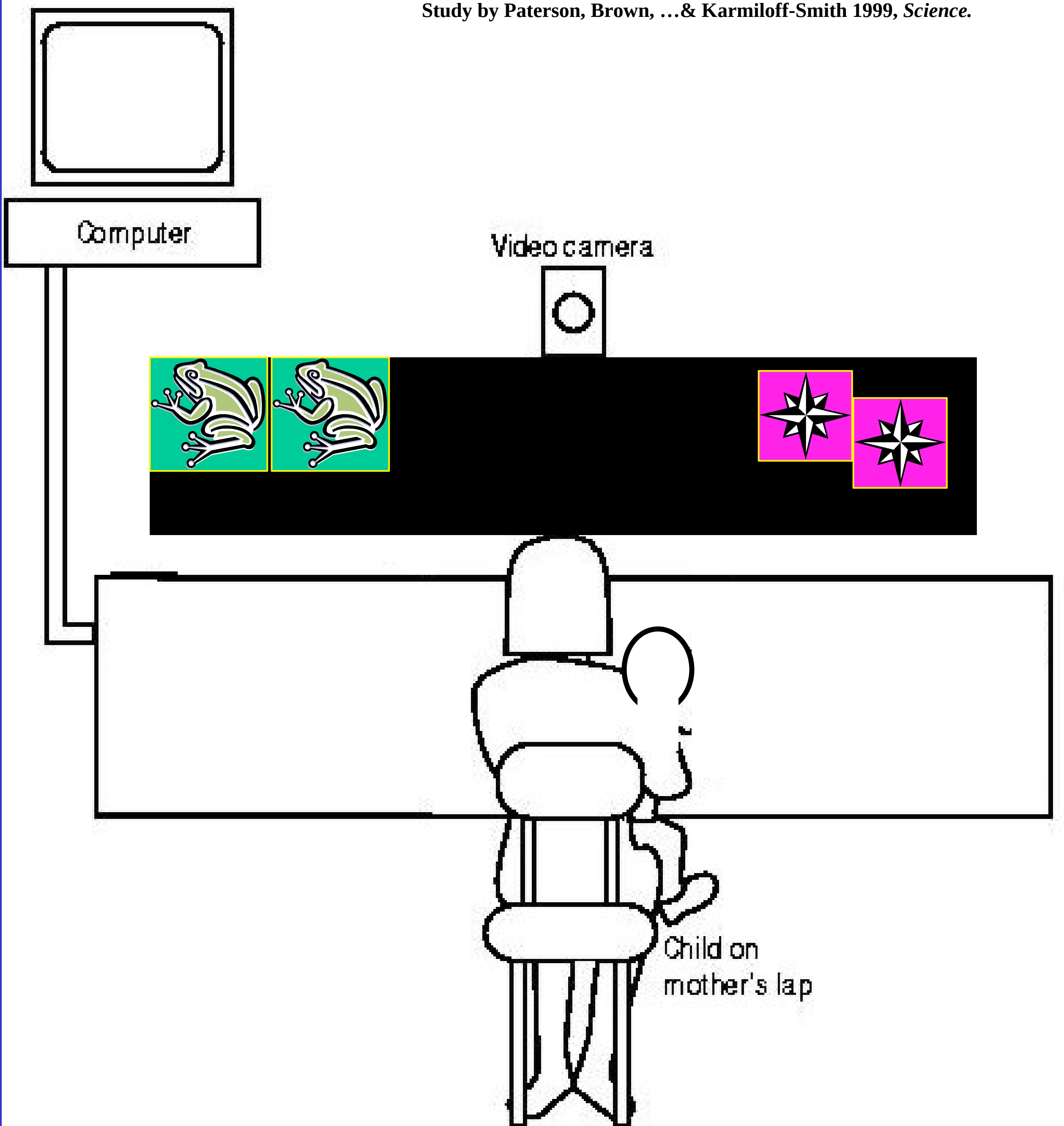
Number development in WS and DS

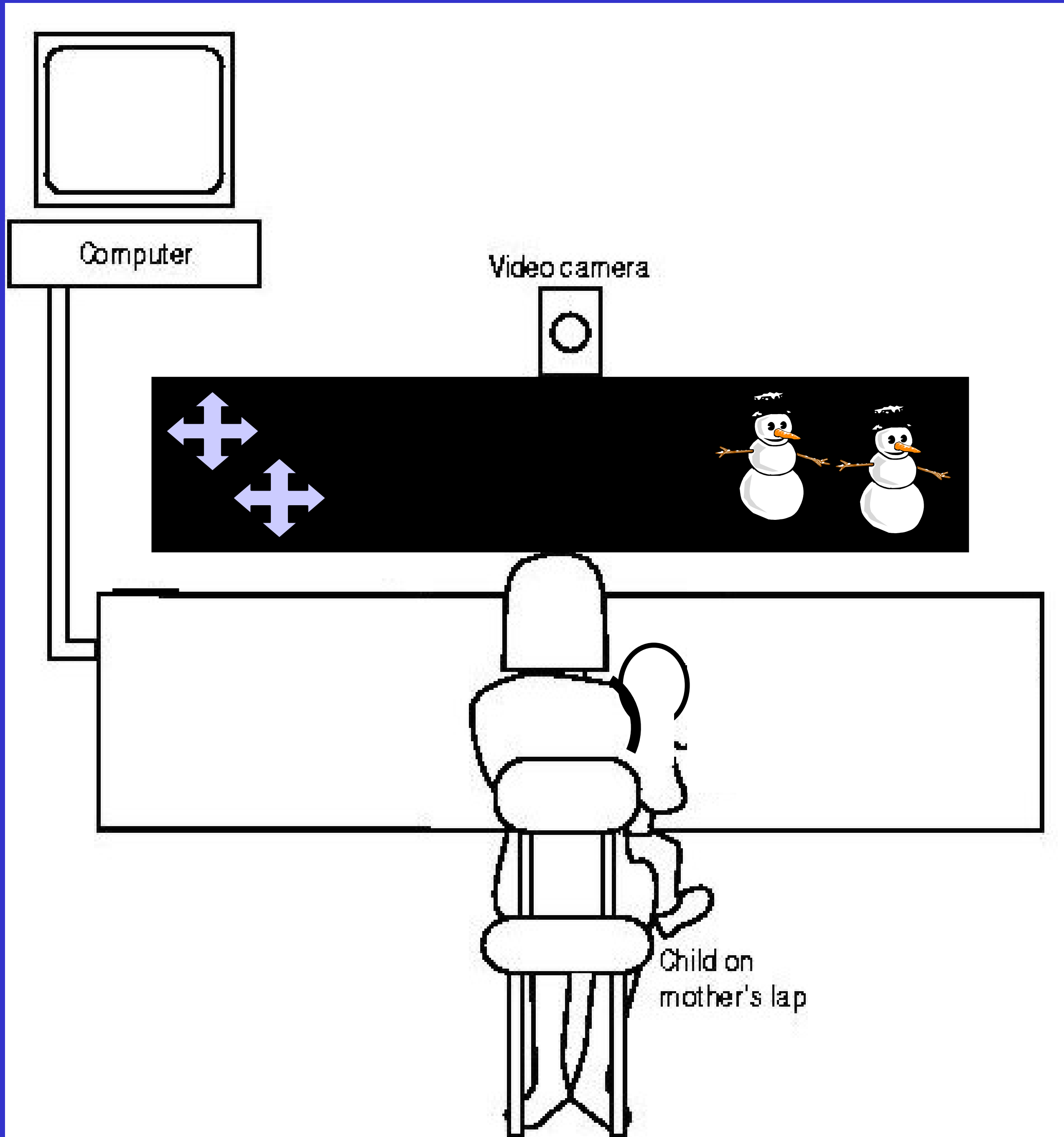

Sarah Paterson

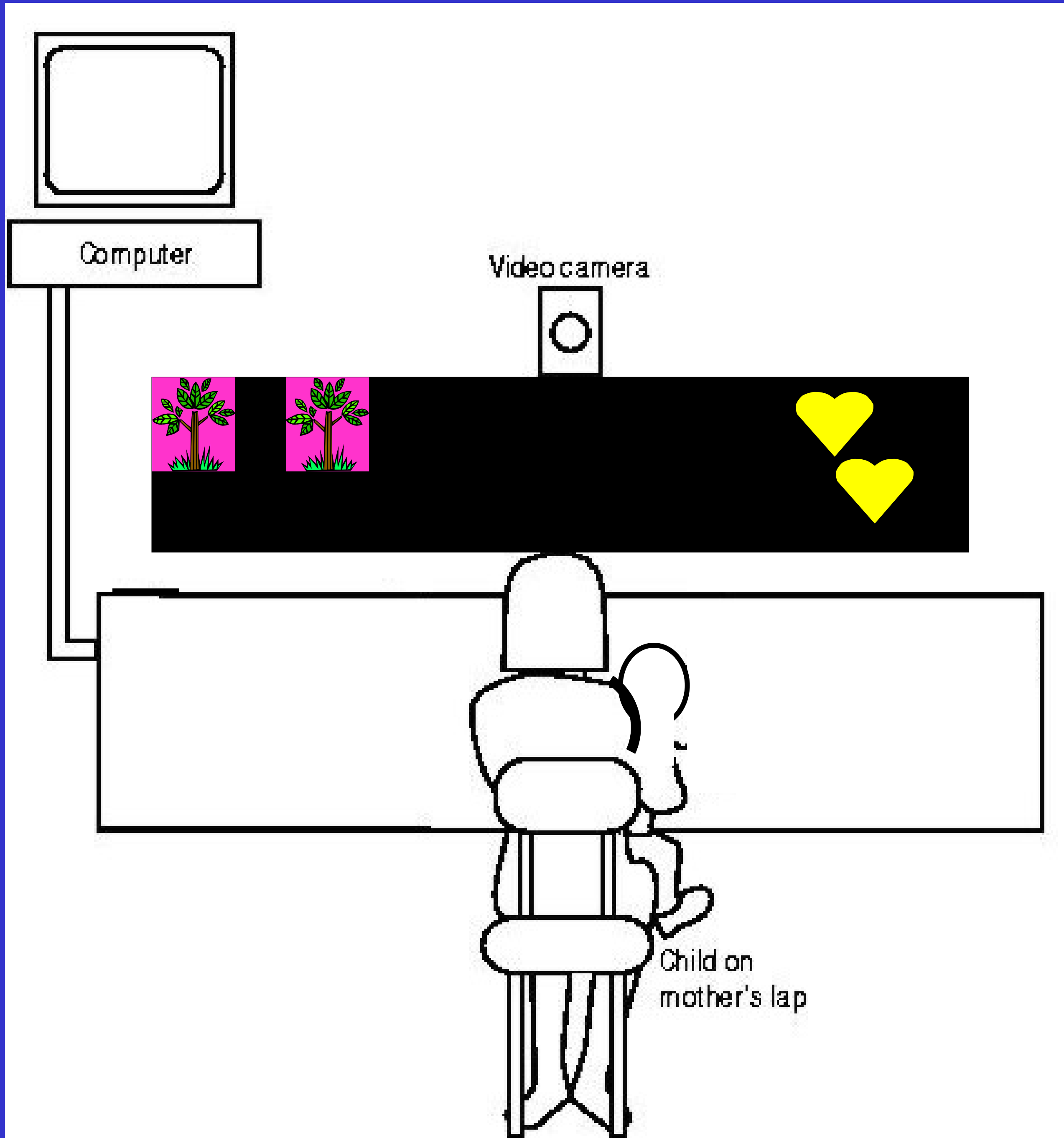
Infancy:

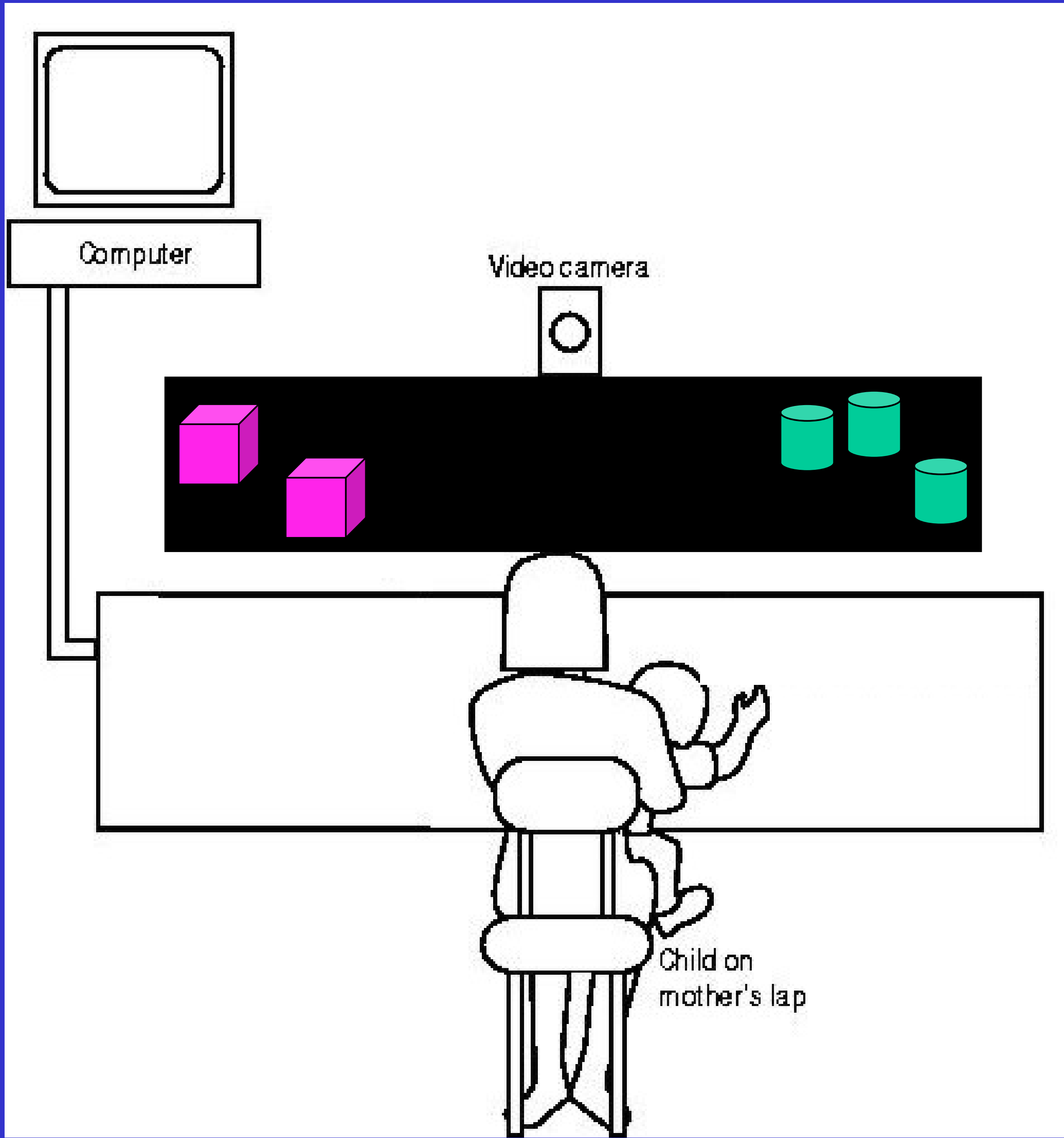
Discrete number discrimination
2 versus 3

Study by Paterson, Brown, …& Karmiloff-Smith 1999, *Science.*




Computer
Video camera
Child on
mother's lap


Computer
Video camera
Child on
mother's lap


Computer
Video camera
Child on
mother's lap

Number development in WS and DS

Infancy:

Exact number discrimination

2 versus 3

WS = CA controls

DS < MA controls

Adolescents and adults:

Butterworth Number Battery

DS > WS on almost all tests

Summary:

infants: WS>DS

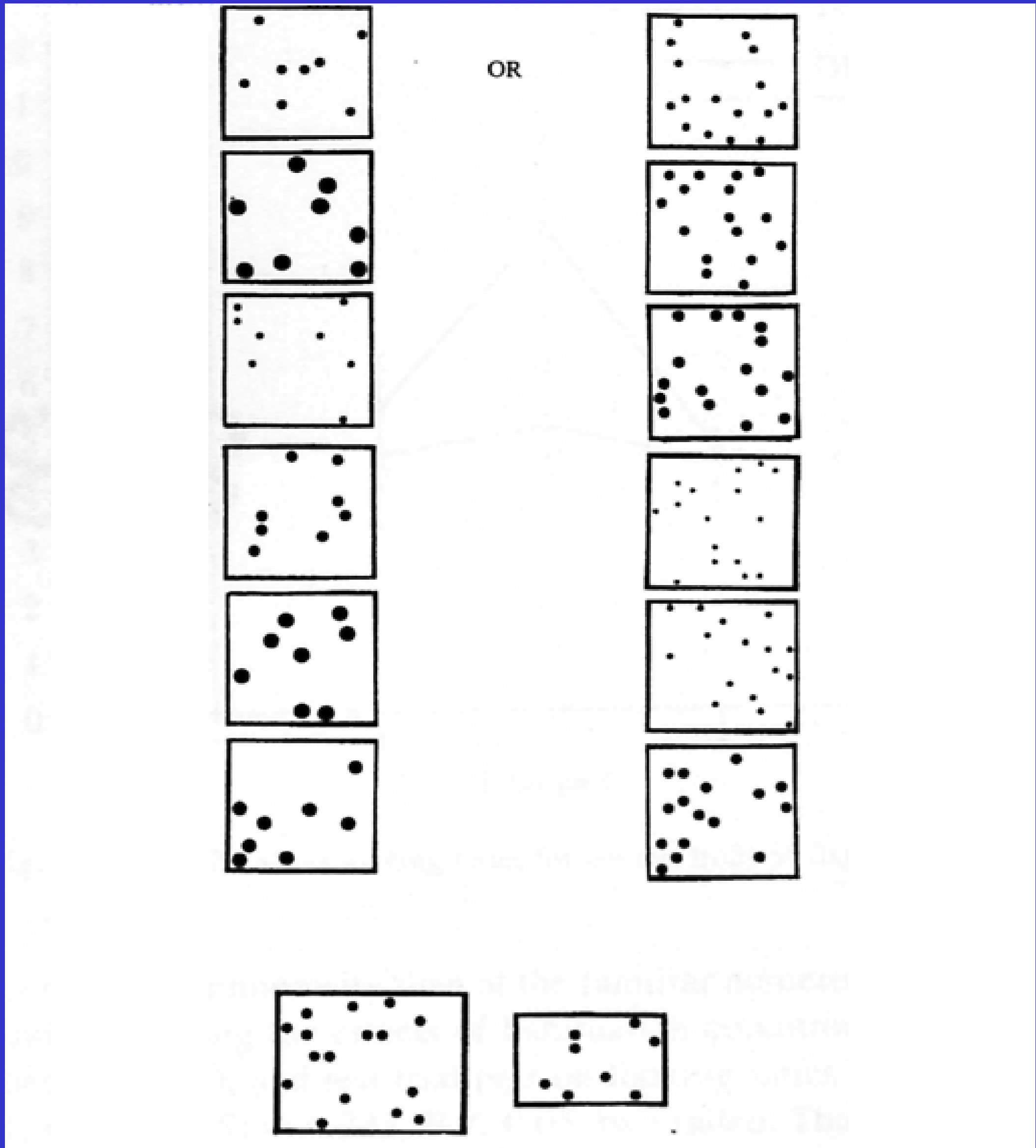
adults: DS>WS

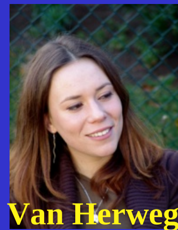
So, success on small exact number isn't necessarily predictive of later number development

exact vs approximate number?

Ratios: 8:12 // 8:16

Study by VanHerwegen, Ansari, Xu & Karmiloff-Smith, in press





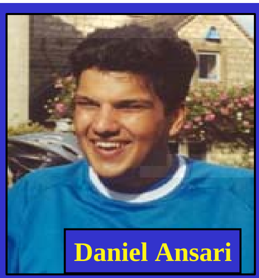
Jo Van Herwegen

Typical infants succeed on 8:16 at 6 months
8:12 at 9 months
Approximate number representations become increasingly precise.

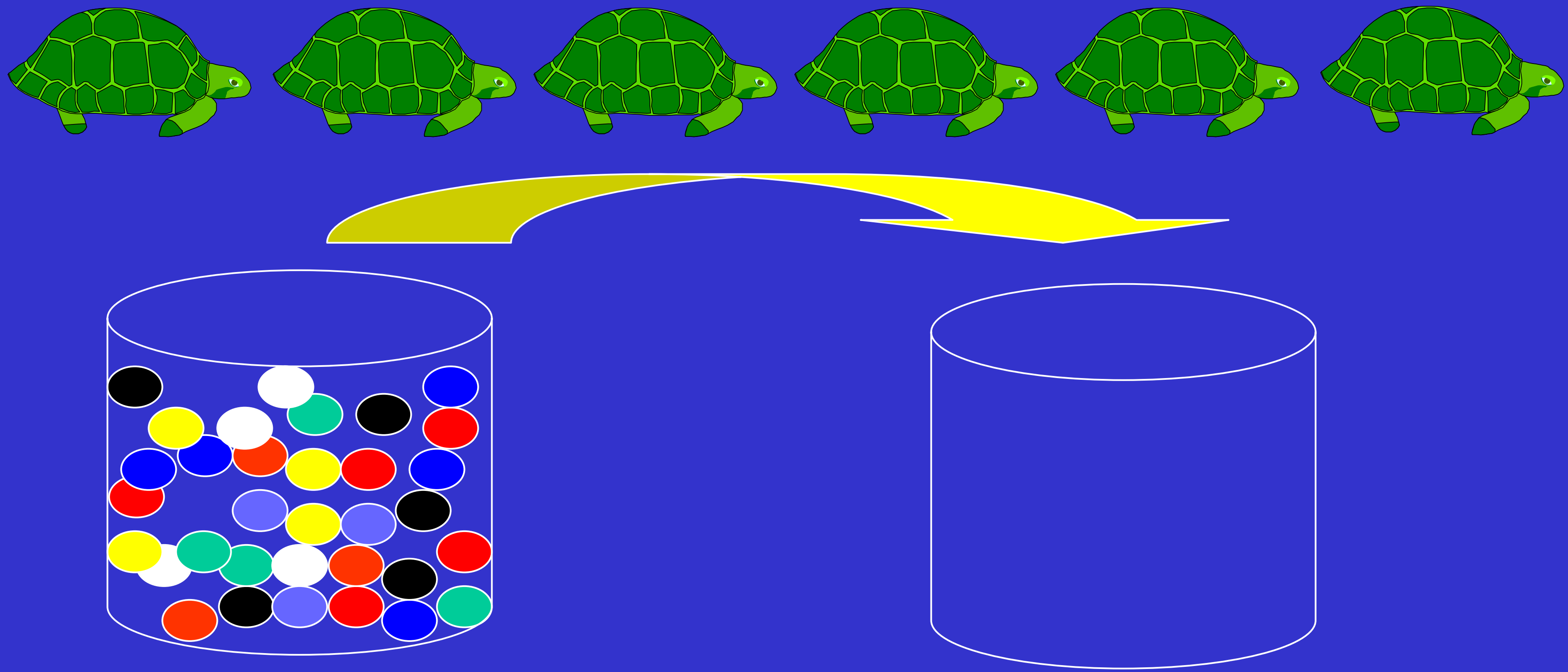
Much older WS children (14-53 months) fail all trials for approximate number

Counting Tasks

for 4-7 year olds



1. Count 1-6 objects – count list: WS: fine



2. "Give-a-number" task - 1-6, 3 repetitions: WS fail

Predicts cardinality? WS=language TD=spatial cognition

Infants: ***Exact*** **small number**

WS = CA controls

DS < MA controls

Approximate **large number**

WS < CA/MA controls

DS? Ongoing: hypothesize DS>WS

Now using eye tracker for both syndromes

Helen Bates MSc Student

Children: Counting:

WS = DS: fine

Cardinality:

WS = fail; Hyothesis: DS>WS

Adolescents and adults:

DS > WS

Individual differences in *approximate* large-number discrimination in infancy = more predictive of later number problems than *exact* small-number discrimination.

Importance of cross-syndrome comparisons to start to build increasingly detailed trajectories of number development
+ ideas for *early* remediation strategies
(e.g. Not counting practice, but rapid large number –magnitude-discrimination practice)

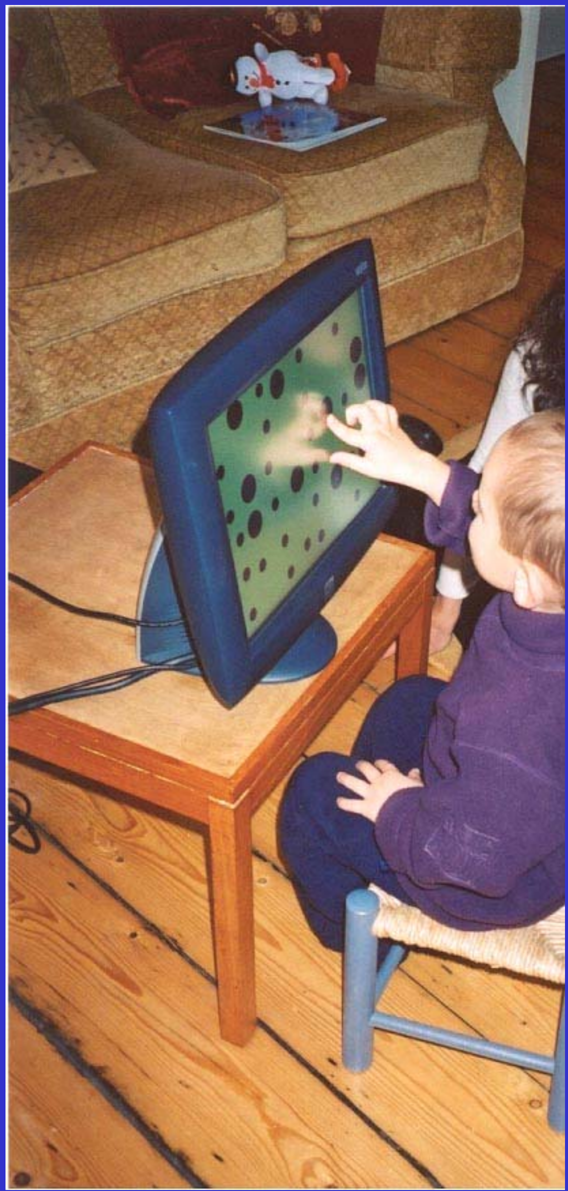
Cross-syndrome studies of attention

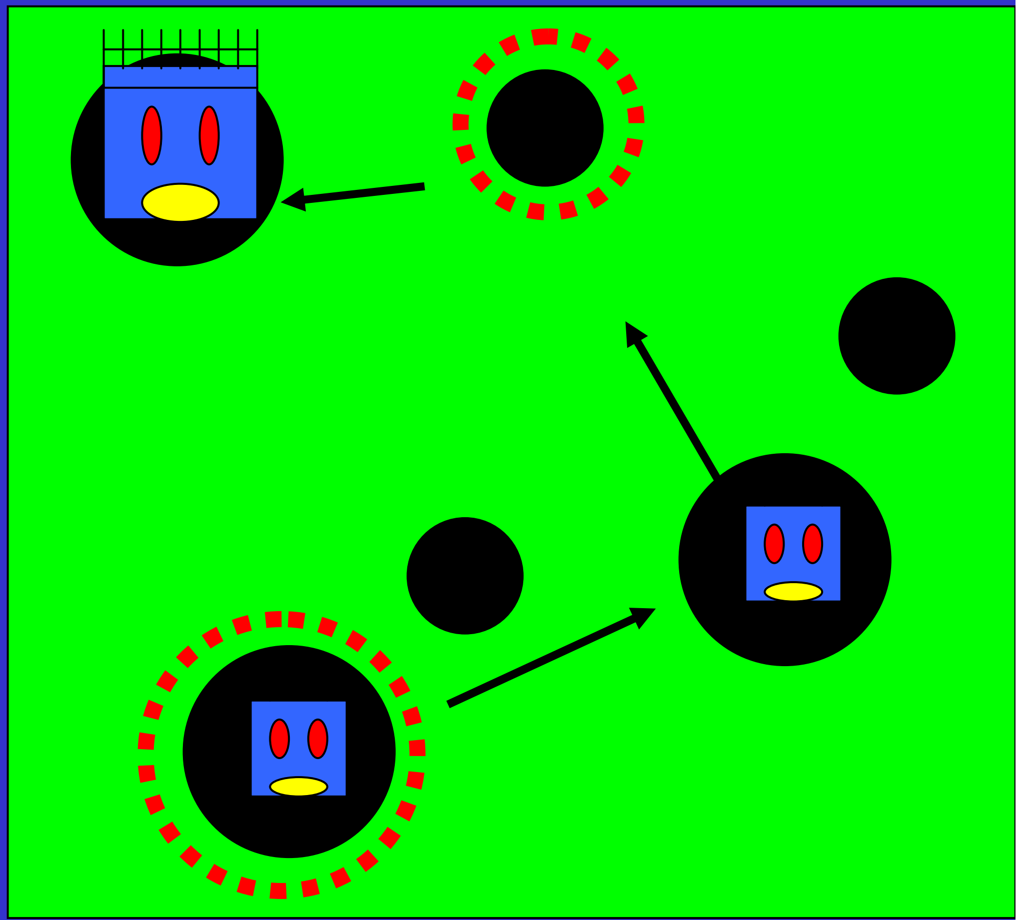
WS/FXS
WS/FXS/DS

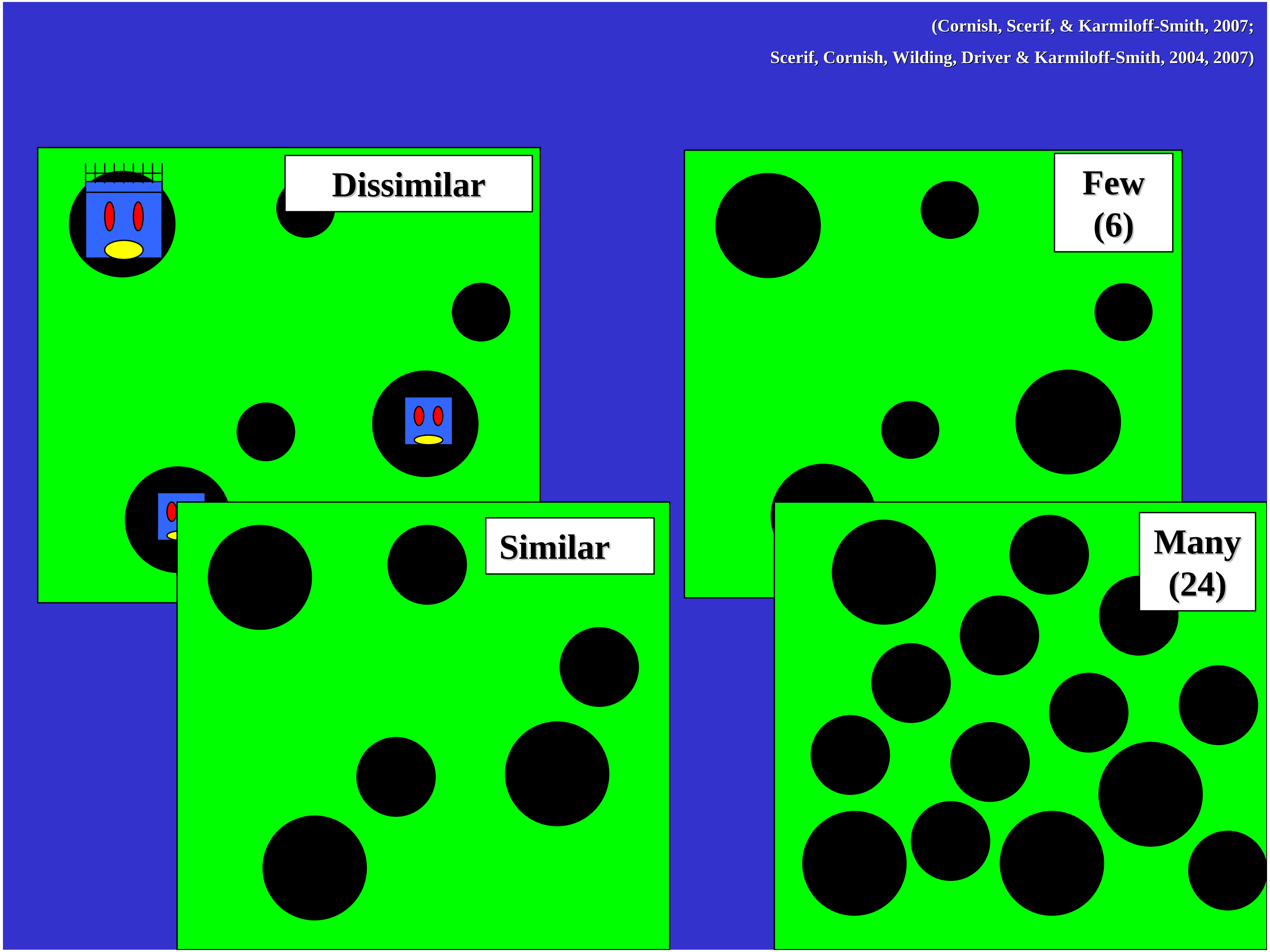
Selective Attention

(Cornish, Scerif, & Karmiloff-Smith, 2007;
Scerif, Cornish, Wilding, Driver & Karmiloff-Smith, 2004, 2007)

Gaia Scerif






(Cornish, Scerif, & Karmiloff-Smith, 2007;
Scerif, Cornish, Wilding, Driver & Karmiloff-Smith, 2004, 2007)
Dissimilar
Similar
Few
(6)
Many
(24)

(Cornish, Scerif, & Karmiloff-Smith, 2007;
Scerif, Cornish, Wilding, Driver & Karmiloff-Smith, 2004, 2007)

Selective Attention

Toddlers and young children with WS, FXS and DS

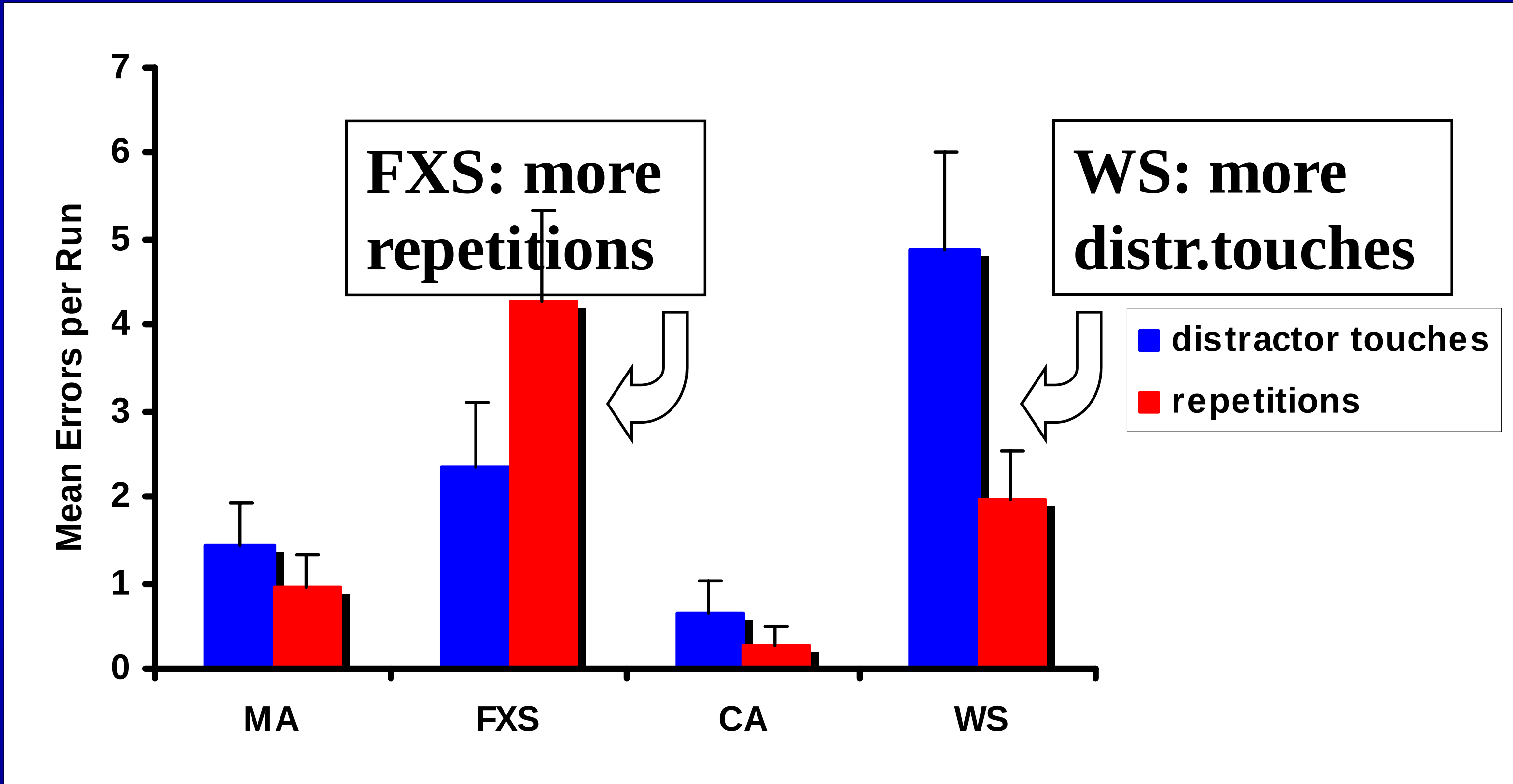
(MA= 18-36 mo., CA = 34-50 mo.)

Search Path: Same all 3 syndromes
Speed & Path: WS = FXS = MA, CA
Search Speed: DS significantly faster than WS

(Cornish, Scerif, & Karmiloff-Smith, 2007; Scerif, Cornish, Wilding, Driver & Karmiloff-Smith, 2004, 2007)

Atypical Errors

MA = 18-36 months, CA = 34-50



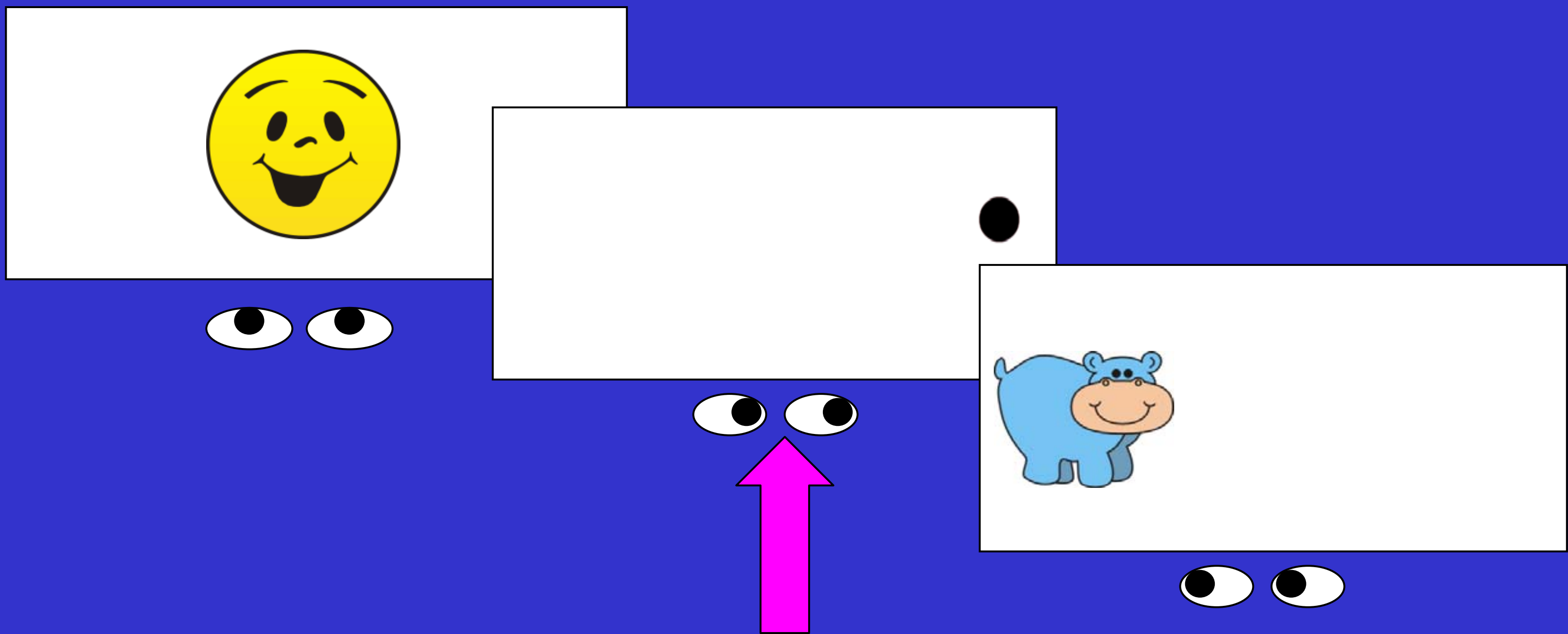
DS: in contrast to the other groups, DS errors did not differ from (younger) MA controls, suggesting delay…but

(Scerif, Karmiloff-Smith, Campos, Elsabbagh, Driver & Cornish, 2005)

Attentional Control of Saccades

Importance of prefrontal cortex for inhibition

Based on studies of patients with PFC lesions (Guitton et al., 1985) and normal 4-month-old infants (Johnson, 1995)



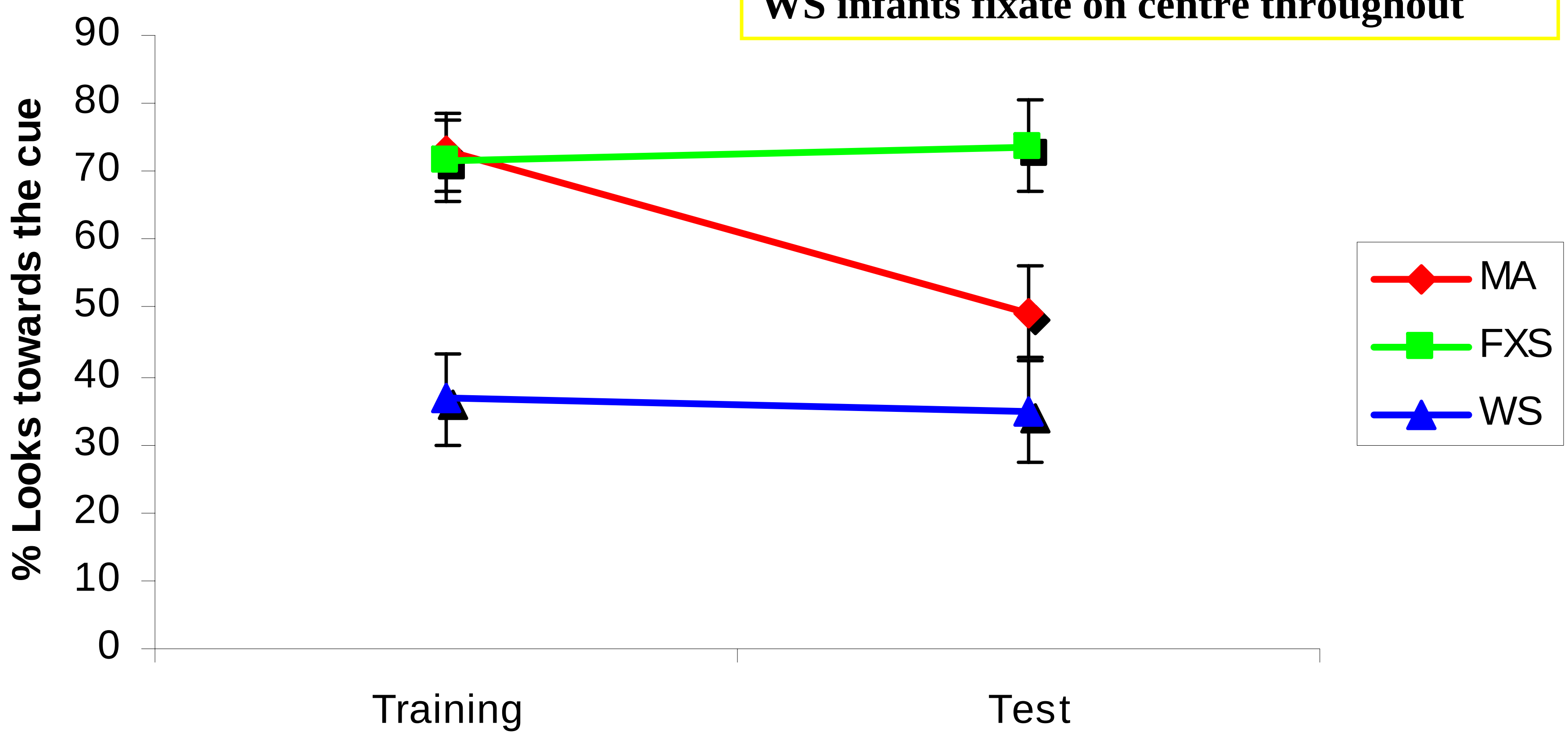
Predictive Cues

Scerif, Karmiloff-Smithet al., 2005, J Cog Neuro)
Cornish, Scerif, & Karmiloff-Smith, Cortex, in press)

9 infants/toddlers with WS, MA = 2-36, CA = 3-41

boys with FXS, MA = 12-30, CA = 14-48

MA controls decrease cue looks
Children with FXS do not
WS infants fixate on centre throughout



(Cornish, Scerif & Karmiloff-Smith, 2007, Cortex)

Simple attentional measures using touch screen and eye-movements = successful in differentiating infants and children with WS from those with FXS

And, recently from those with DS

Ongoing: longitudinal studies of all 3 syndromes across all aspects of attention

Justin Cowan, Postdoc

Anne Steele, PhD student

Cross-syndrome studies of face processing

Different labs worldwide (Benton/Rivermead):

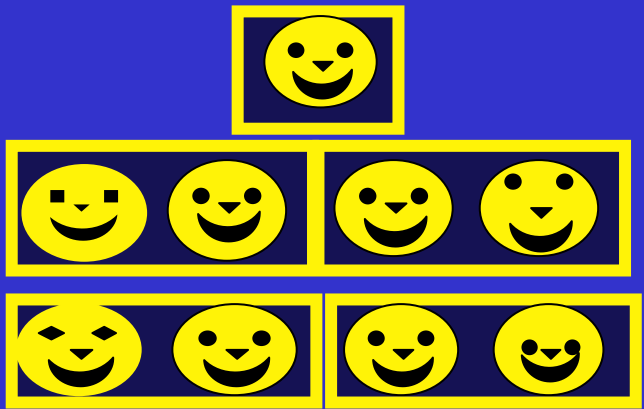
WS good at face processing: *‘in the normal range’*

Dagmara Annaz

Michael Thomas

Kate Humphries

Spared face processing module in WS?



Inversion effect (hallmark of *configural* processing)

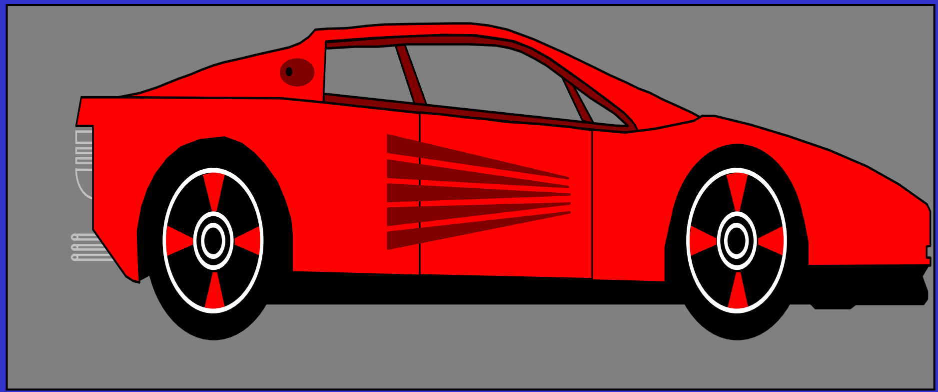
doesn’t **emerge developmentally in WS.**

Karmiloff-Smith, et al., 2004

What about WS brain?

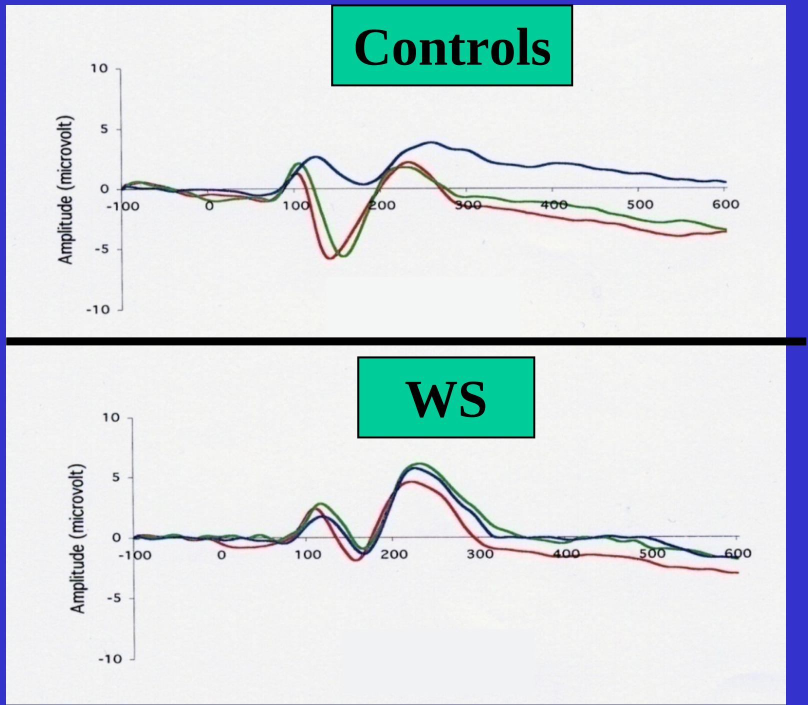
Sarah Grice
Gergo Csibra
Mark Johnson
Michelle de Haan

WS adolescent in Geodesic HD-ERP net



Grice et al., 2001, 2003

Normal modularisation of function, and WS lack thereof

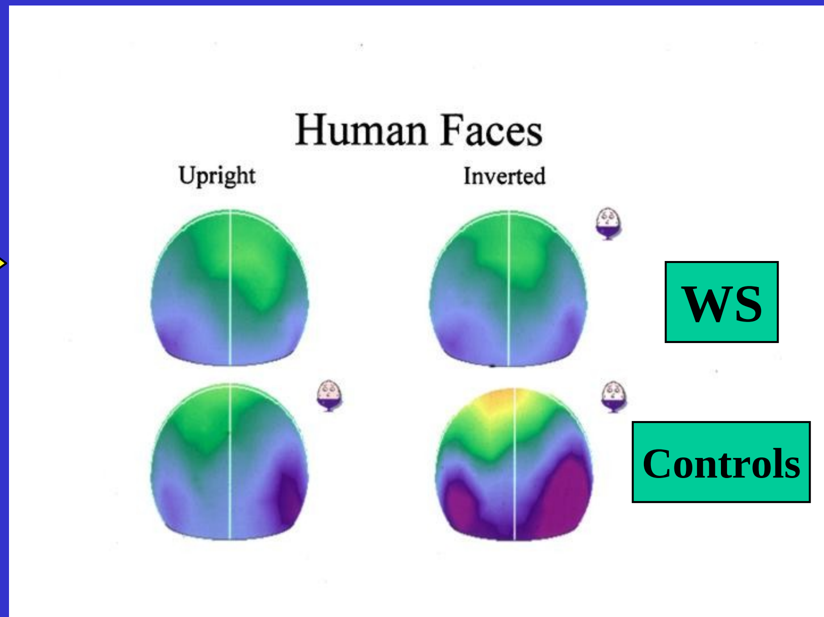


Healthy controls:

Progressive restriction of input type

WS: failure to specialise

WS: failure to localise

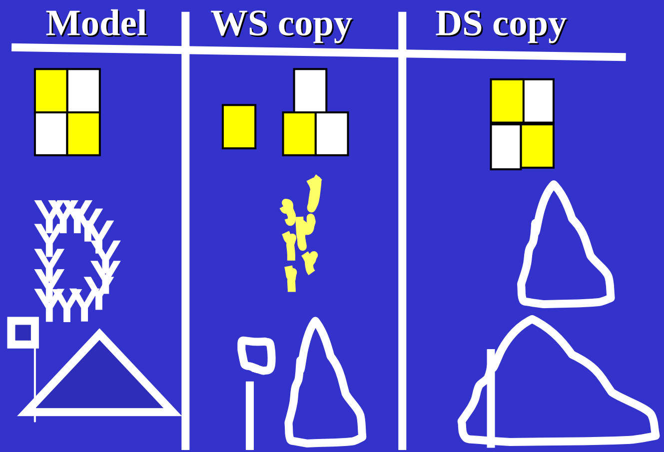


Healthy controls:
Progressive restriction of brain localisation

WS: not only faces

Face processing:
Space processing:
Sound processing:

) WS: all processed more featurally
) than configurally
) DS: more holistic/global
) than featural



Note change-Y
Contour change-N

Mayada Elsabbagh

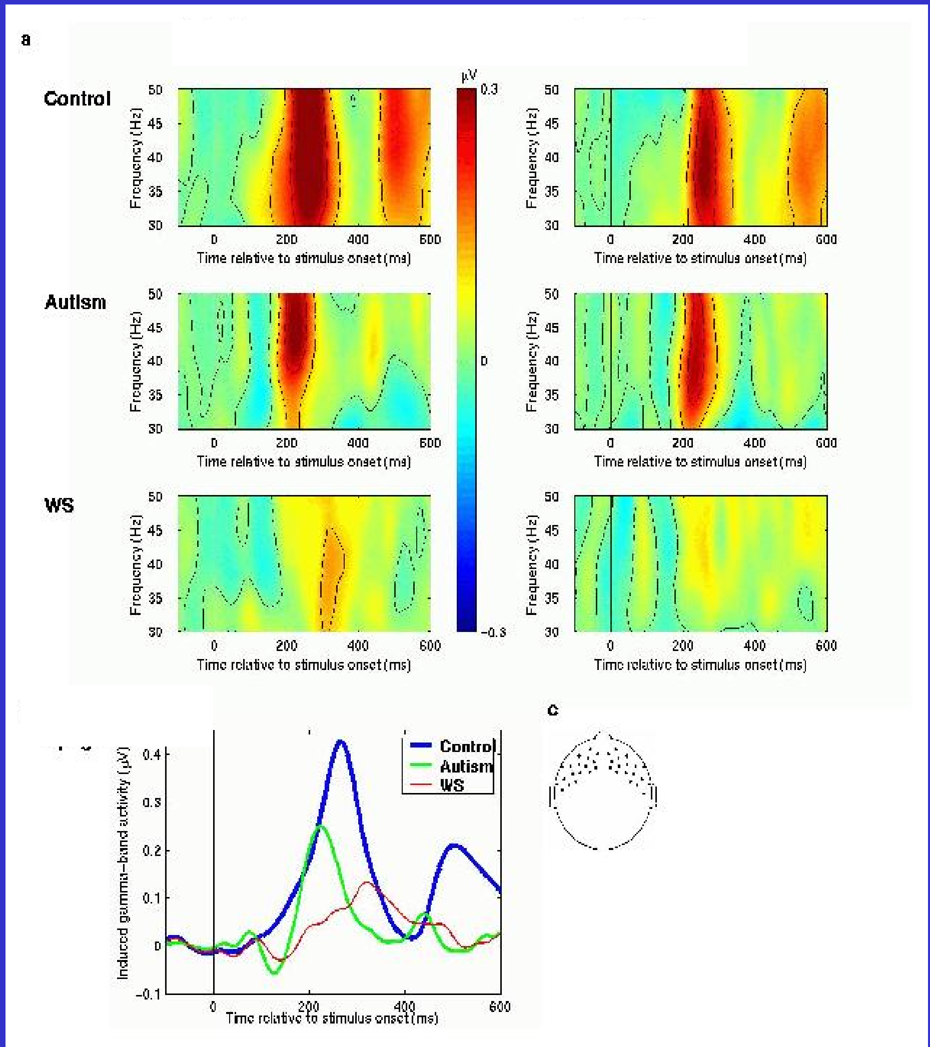
WS: Failure to bind features into a whole

Both WS and autism:

“featural” processors…..

Gamma-band bursts in WS and autism: integration/binding of features

Sarah Grice
Gergo Csibra
Mark Johnson
Michelle de Haan



Atypical brain function in both syndromes,
but cross-syndrome brain difference

Rethink notion of "featural at cognitive level…..

Ongoing research:
binding patterns in DS and FXS?

Cross-syndrome studies of communication

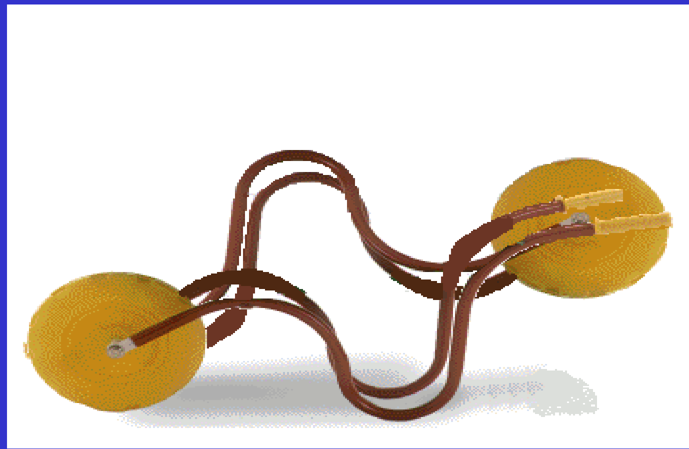
Preferential looking

“Look! Look at the chair”

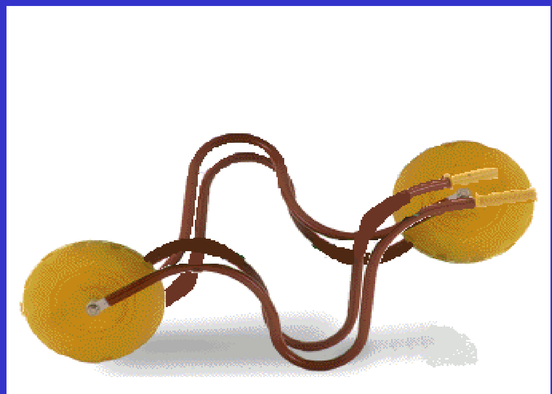
Learning new words

Thierry Nazzi

Sarah Paterson



“ Look, look at the blicket”

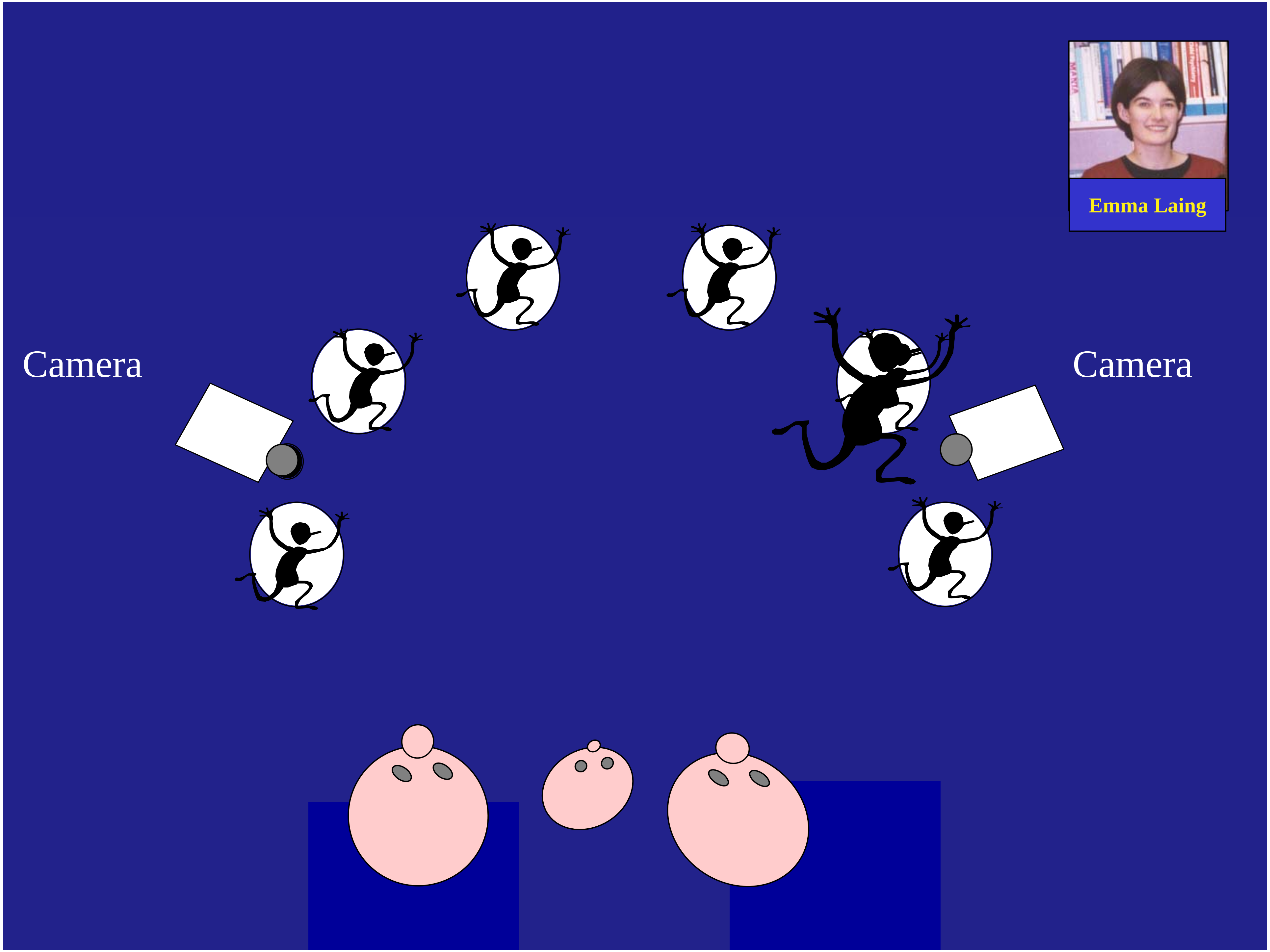


“Where’s the blicket?”

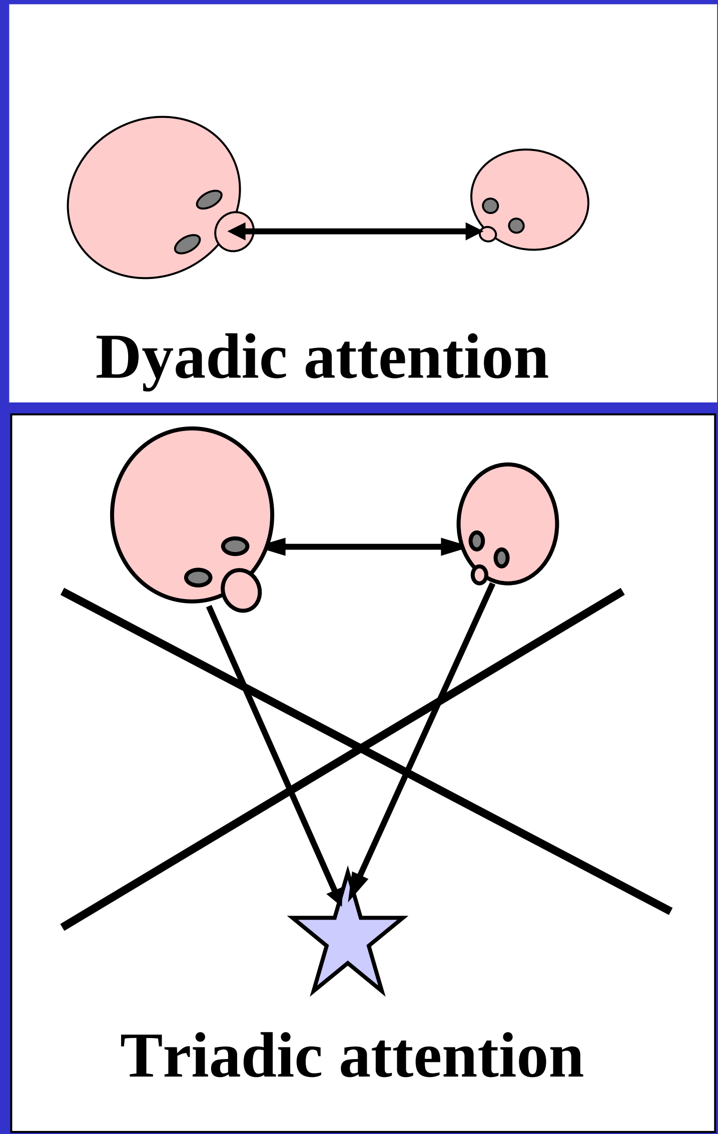
Summary infant word comprehension & word learning

- **WS and DS *both* delayed *to same degree*, despite huge language differences in adulthood: WS adults outstrip DS adults**

- **Like number, infant language start-state profiles DO NOT always resemble adult phenotypic end-state**


Emma Laing
Camera
Camera

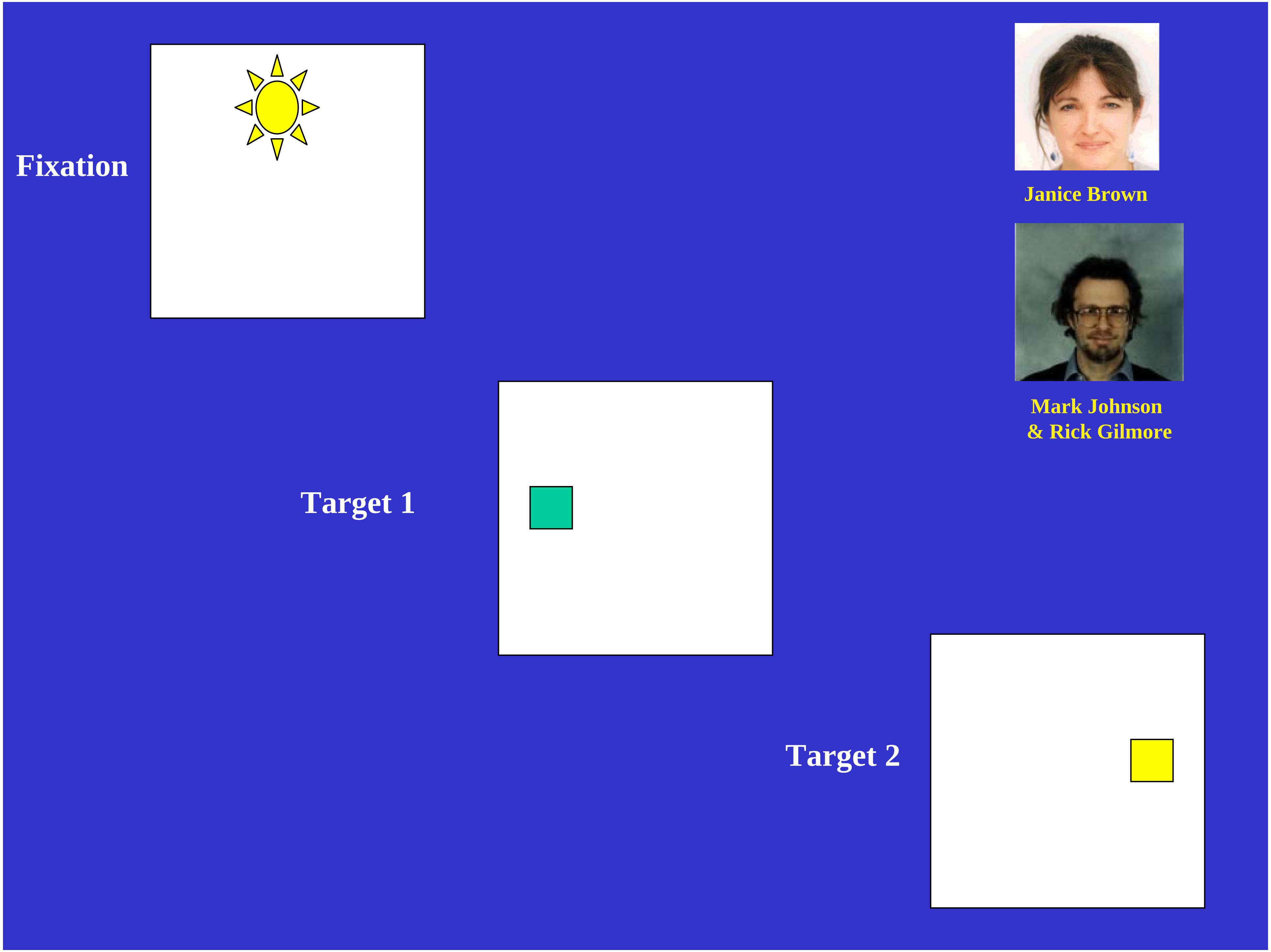
WS
Joint Attention



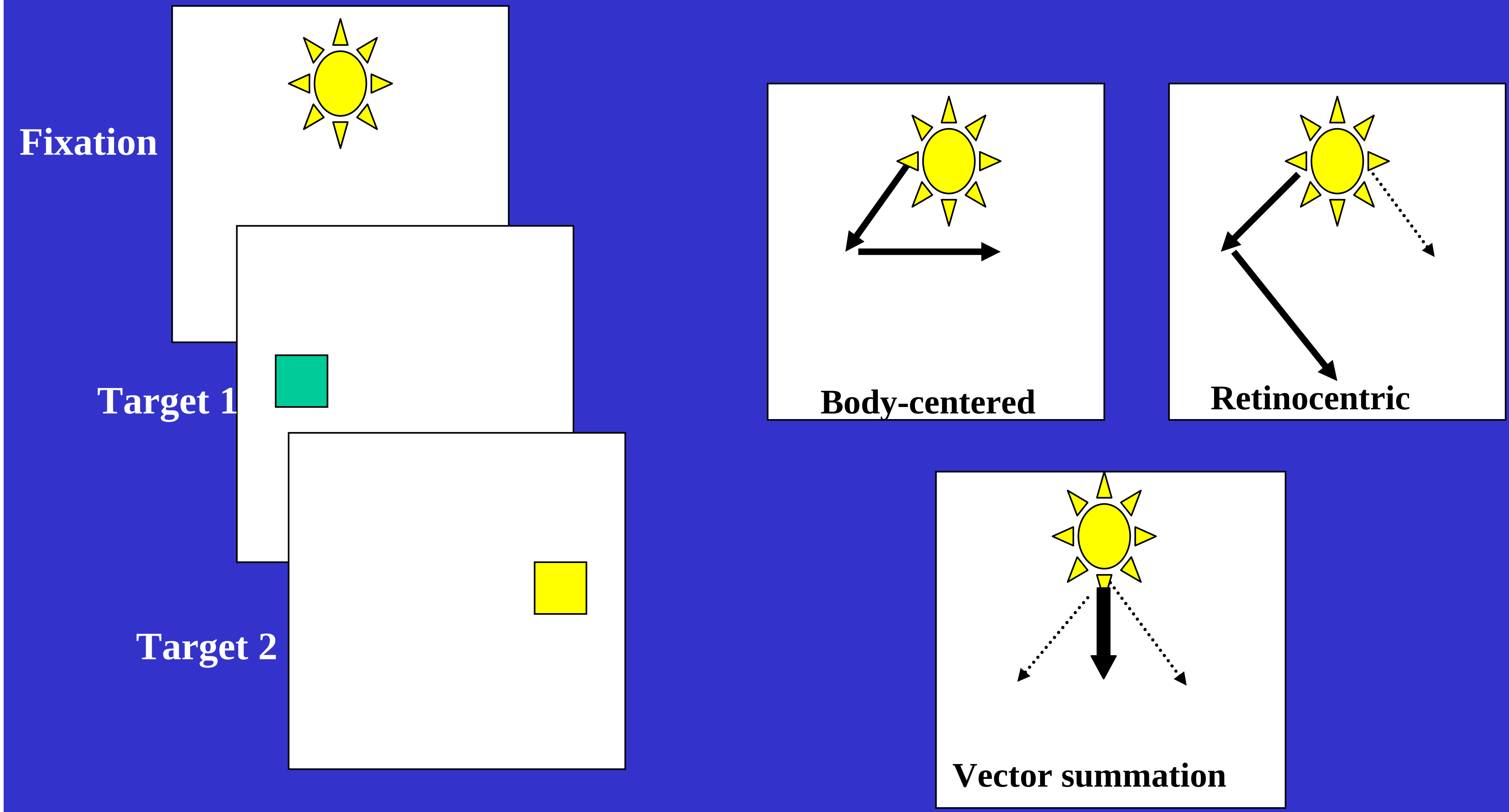
Does same cause explain DS delay?

Is explanation of late language confined to speech/language/communication?

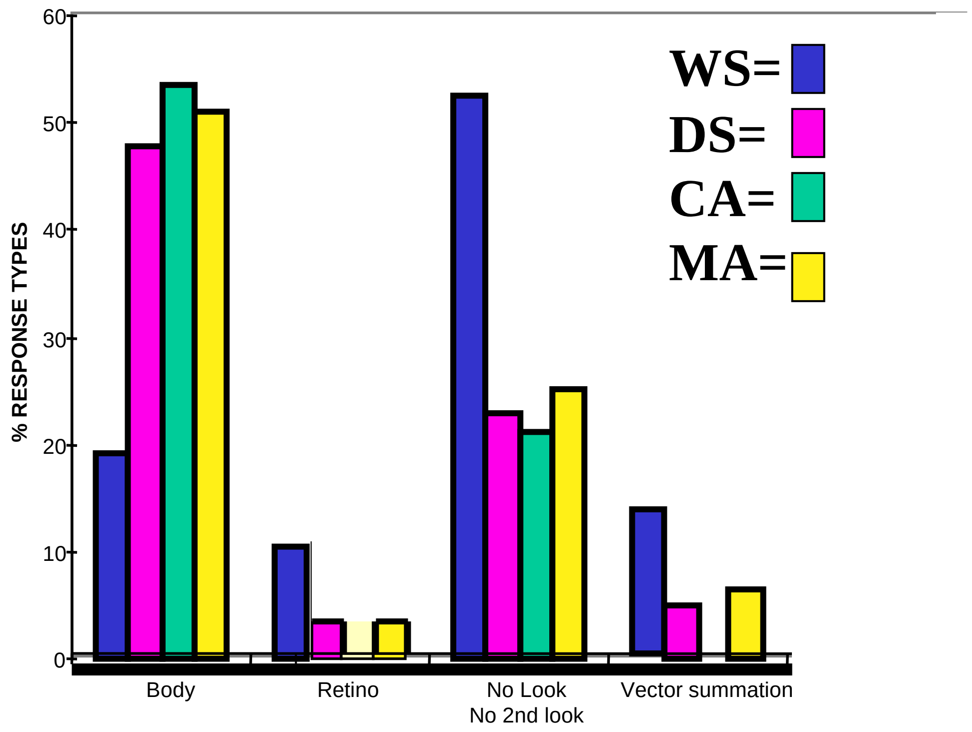
within *same* domain?


Fixation
Target 1
Target 2
Janice Brown
Mark Johnson
& Rick Gilmore

Visuo-spatial precursors to socio-communication in typical & DS infants and toddlers, impaired in WS



Brown et al., 2003



DS/WS = *different* causes for *similar* initial language delay

Basic deficits in visual system *early*

in WS developmental trajectory

- **sticky fixation**
- **focus on featural detail:**

cascading developmental effects over time

across several *emerging* higher-level

linguistic/cognitive systems:

number, attention, faces, language.

In summary

√Full developmental trajectories:

cross-sectional and longitudinal

√ In-depth, detailed *cross-domain* cognitive-level studies to plan intervention (visual->languge)

√ In-depth *cross-syndrome* studies:

uncover more subtle differences

(not just X = spared/Y = impaired)

with syndrome-specific implications for early remediation

√ Different behavioural & brain imaging measures on *same individuals*, across developmental time

EYE-TRACKER, MRI, fMRI, MEG, PET, HD-ERP, MRS, NIRS, etc.

Thank you!
Merci!

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http://www.psyc.bbk.ac.uk/research/DNL/personalpages/annette.html