

A Contextual Neuropsychological Approach to Neurodevelopmental Disorders

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We see:

prevalent syndromes, e.g.,
Noonan/Rasopathies, 22q11.2DS,
PWS, Klinefelter;

And often:

rare syndromes, such as Kleefstra,
16p11.2, KBG, KoolenDeVries,
Kabuki, SATB2 Associated Syndrome
(SAS).

www.cvneuropsychiatrie.nl

www.vvgi.nl



Our clinical (research) strategy for a specific syndrome:

- For individual purposes: Detailed assessment of cognition
- Establishing individual profile of cognitive ‘action in context’
- Matching or developing training/treatment and guidance strategies

AND WHEN SEEING MANY MORE PATIENTS:

- Cumulative (group) data → possible establishment of cognitive phenotype



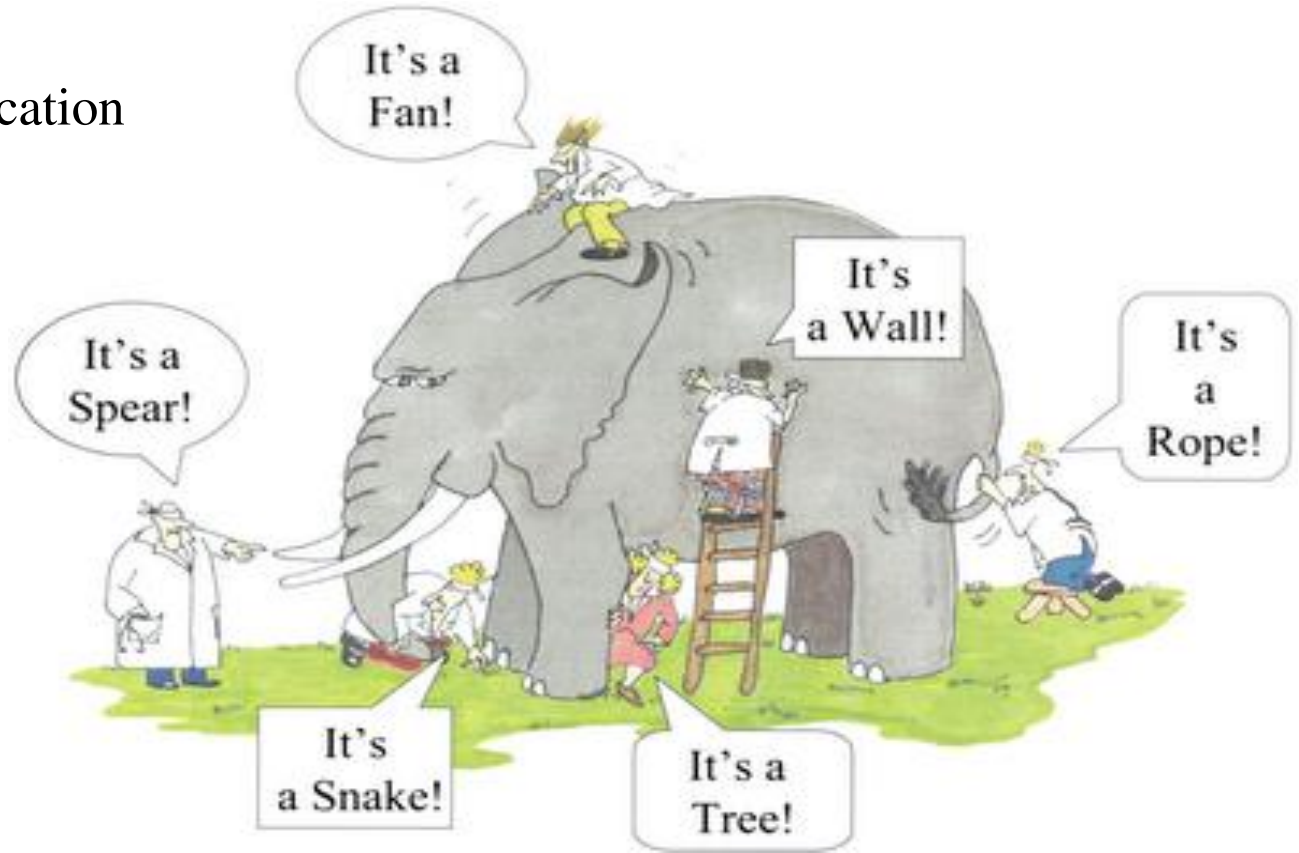
Outline of the talk

- Contextual neuropsychology: A clinical strategy accompanied by research
- Neuropsychological framework: Key concepts relevant for description and understanding of (problems in) daily functioning
- Social information processing/social cognition: Description and example of a genetic disorders in which fruitful discovery of a syndrome-specific profile and training was made
- The case of PMS: Regression, cognition and behaviour. Adaptation and further development of assessment and training methods



Same gene – different appearances: Why? What now?

Clinical classification
problem

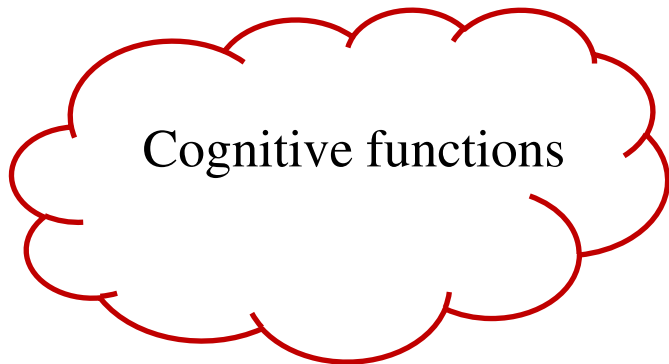




Neuropsychological framework

Neuropsychology

- clinical research discipline that combines neurosciences with social sciences
- relations between brain and human cognitive, emotional, and behavioural functions
- cognitive phenotyping



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Mental processes involved
in thinking and experiencing



Neuropsychological framework



- intelligence
- language
- perceptual and motor functions
- attention and speed of information processing
- executive functions
- learning and memory
- affective information processing, social cognition



You CAN perform neuropsychological tests in children with moderate to severe ID, even when nonverbal.

Vermeulen, K., Staal, W., Janzing, J., Van de Ven, F., Oomens, W., Van Dongen, L., Van Bokhoven, H., Kleefstra, T., Egger, J. Computerized Neurocognitive Assessment in Rare Genetic Disorders with Moderate to Profound Intellectual Disabilities: A proof of principle study. *Clinical Neuropsychiatry*, 2018.

- Tasks from the (adapted) Cambridge Automated Neuropsychological Battery (CANTAB) can be used from a developmental age of 2,5 years
- Can also be used transspecies



Neuropsychology in PMS

Genes, Brain
and Behavior

Official publication of the International Behavioural and Neural Genetics Society

Genes, Brain and Behavior (2016)

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Neuropsychological phenotype and psychopathology in seven adult patients with Phelan-McDermid syndrome: implications for treatment strategy

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emotional sequelae. It is concluded that the treatment of 22q13.3-associated psychopathology should include prescription of mood-stabilizing agents in combination with individually tailored contextual neuropsychological measures.

Keywords: Atypical bipolar disorder, cerebellum, cognition, contextual neuropsychology, mood-stabilizing treatment, neuropsychological phenotype, Phelan-McDermid syndrome, SHANK3

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- moderate to severe intellectual disability with profound
- communication deficits, characterized by dysfunctions in
- speech/language and
- executive attention, as well as
- cognitive alexithymia and
- emotional instability



Treating impairments in social cognition

- Behavioural training and management
- Psycho-educational interventions
- Cognitive Behavioural Therapy (CBT)
- Coaching, psychosocial support

- Targeted to multimodal therapies such as SENS (Roelofs et al. 2018)





What to do?

Some recommendations from a neuropsychological viewpoint

Integrating neuropsychological diagnosis and treatment into the care for individuals with PMS by annual neurocognitive and behavioural screenings until adulthood and beyond

Conducting neuropsychological assessments at key transitional stages (e.g., 3y; 10-12y; 16-18y)

Adapt evidence based interventions for cognitive, behavioural or psychosocial problems from other populations to meet the needs of individuals with PMS (e.g., DBT)

Further development of reliable and valid proxy based assessment instruments for neurocognitive functioning

Exploring the utility of automated (augmentative) communication devices to enhance communication and training and guidance possibilities

(Possibly) Exploring the utility of Transcranial Magnetic Stimulation based on studies about cerebellar basic symmetrization / regulation of motor function, cognition and behaviour

GRACIAS!

Thanks for your (sustained) attention!

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