

Why we monitor the cognitive development of children and adolescents with 22q11DS

*A study by the International 22q11.2 Deletion Syndrome
Brain Behavior Consortium (IBBC 22q11DS)*

Sasja Duijff, PhD



Universitair Medisch Centrum
Utrecht

- The average child with 22q11DS
- Schizophrenia
- Cognitive development & schizophrenia
- IBBC study
- Recommendations



The average child with 22q11DS



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DOES NOT EXIST!!!!

...yet there are similarities...



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(Common) personality characteristics in 22q11DS



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- Sweet
- Creative
- Determined
- Loyal and focussed
- Pleasant
- Musical
- Love to chat

(Common) personality characteristics of parents with a child with 22q11DS



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- Committed
- Patient
- Determined
- Loving
- Powerful
- Creative

(Common) cognitive characteristics in 22q11DS

- Nearly 100% learning difficulties
 - Difficulties with comprehension
 - Word finding difficulties
 - Difficulties with perceptual organization
-
- Relatively strong short term memory
 - Relatively strong processing speed
 - Verbally strong impression

Confusing profile for parents, teacher & child

22q11DS is an invisible handicap

(Common) behavioral characteristics in 22q11DS



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- Fearful at a young age
- Social behavior:
 - prefer older or younger children
 - few friends, socially isolated
- Social clumsiness
- Attention and thought problems
- Strong moodswings
- Increased risk for psychiatric problems:
ADHD/ ASD/ schizophrenia

Schizophrenia



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- Affects 25-30% of adolescents with 22q11DS
- One of the major concerns of parents, therefore focus of attention by researchers

Schizophrenia

- Is *not* a split-brain condition
- Has *nothing* to do with multiple personalities

Schizophrenia - DSM 5



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A. Characteristic symptoms:

Two (or more) of the following (at least 1 of these should include 1-3)

1. Delusions
2. Hallucinations
3. Disorganized speech
4. Catatonia
5. Negative symptoms, i.e. restricted affect or avolition/asociality

B. Social/occupational dysfunction

C. Duration: continuous signs of the disturbance persist for at least 6 months.



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PSYCHIATRIE.

EIN LEHRBUCH

FÜR

STUDIRENDE UND AERZTE

VON

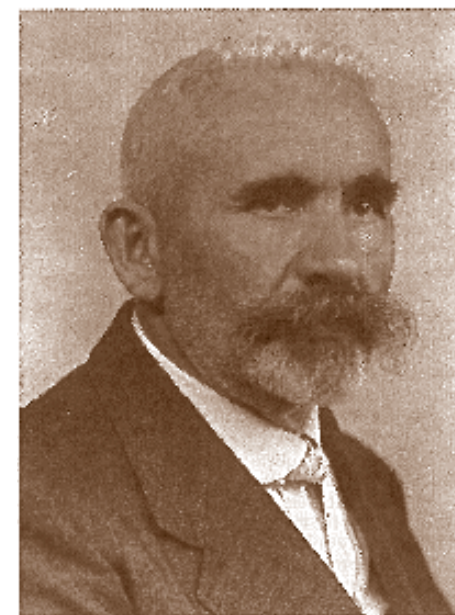
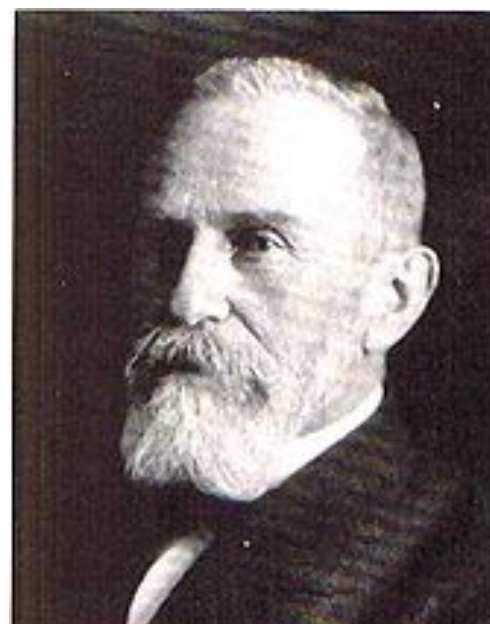
Dr. EMIL KRAEPELIN,
PROFESSOR AN DER UNIVERSITÄT HEIDELBERG.

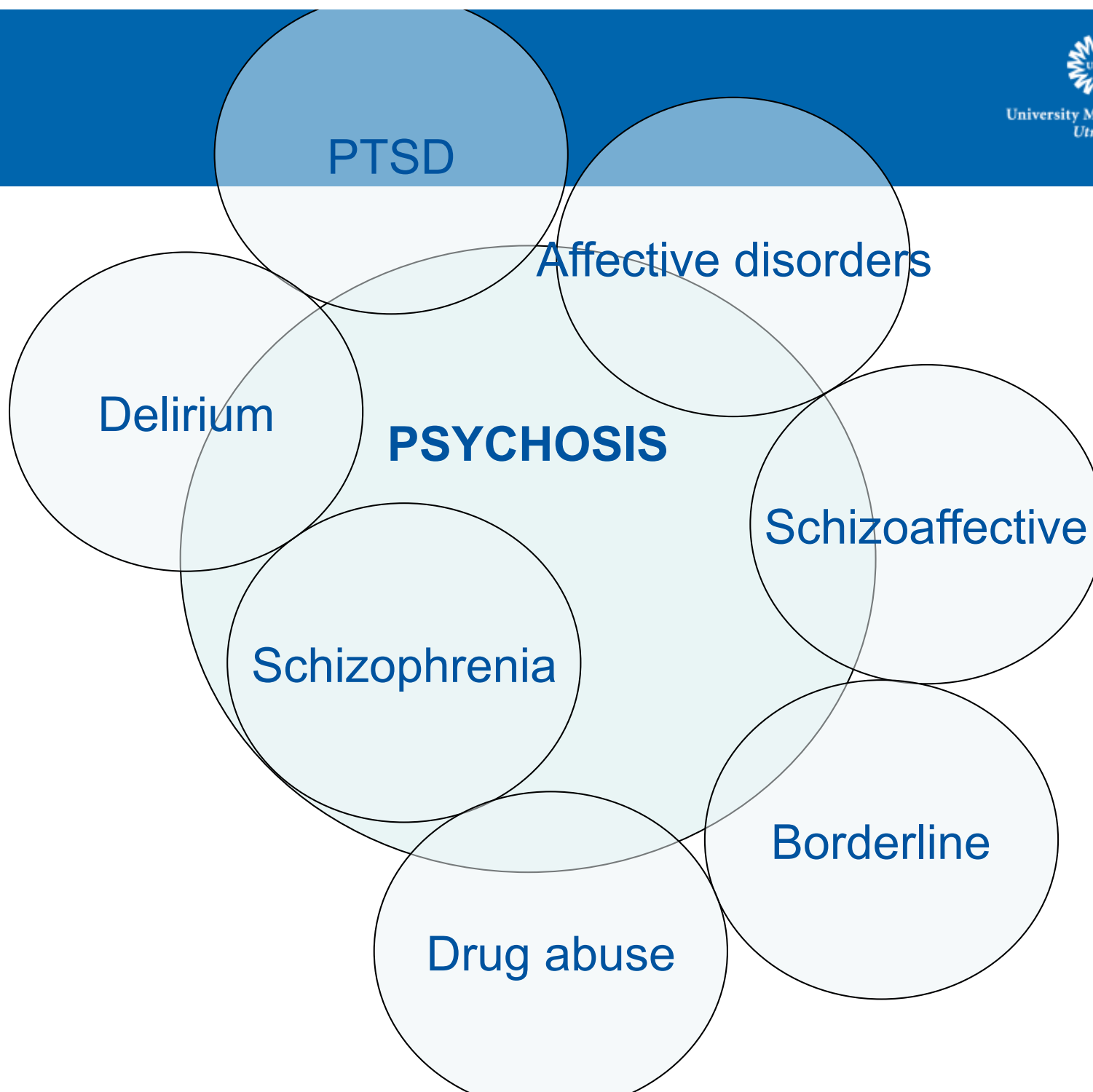
SECHSTE, VOLLSTÄNDIG UMGEARBEITETE AUFLAGE.

I. BAND.
ALLGEMEINE PSYCHIATRIE.



LEIPZIG,
VERLAG VON JOHANN AMBROSIOUS BARTHEL
1899.

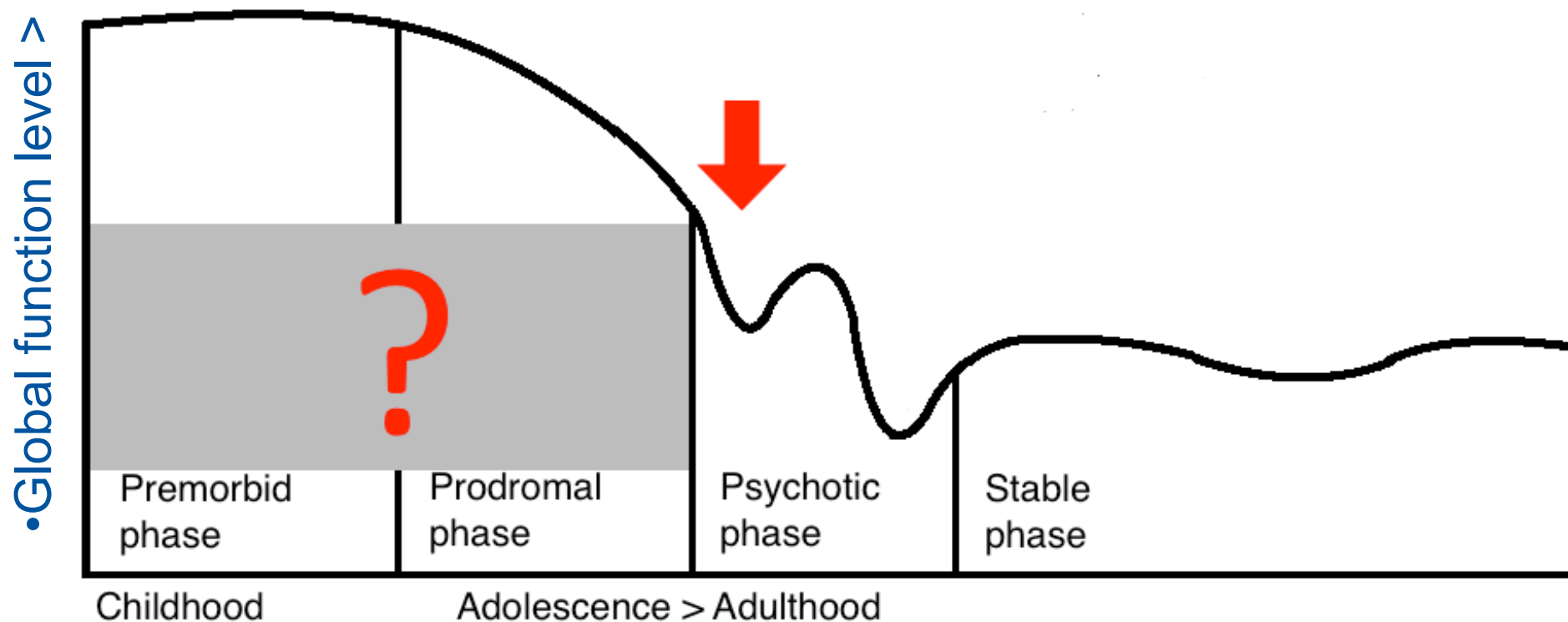




Schizophrenia trajectory



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22q11DS



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Summary findings so far



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Based on previous cognitive research:

- Overall intelligence level does not appear to be *strongly* associated with global *childhood* psychopathology (ASD/ ADHD) in 22q11DS
- Individuals with psychotic disorders appear to have lower (verbal) IQ
- All cross-sectional data; what about cognitive development?

Cognitive development & schizophrenia in 22q11DS



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•5.5 years

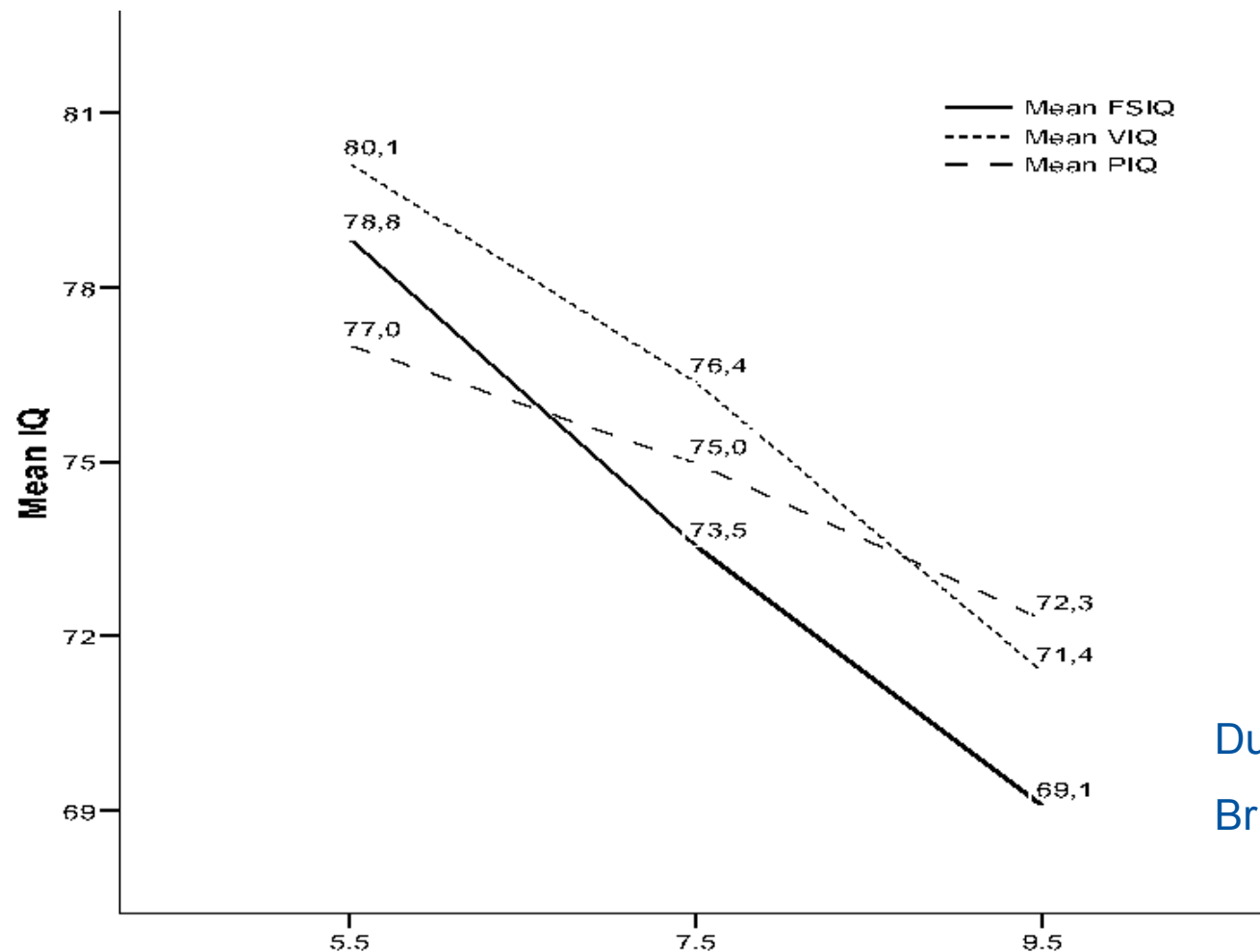
WPSSI-R / SON

•7.5 years

WISC-III

•9.5 years

WISC-III



Duijff et al.

Br J Psychiatry, 2012

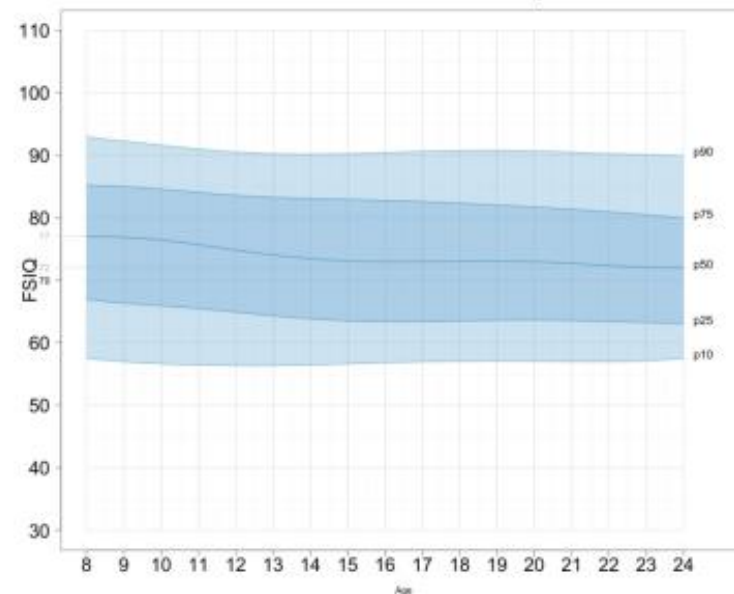
- International Brain Behavior Consortium on 22q11.2 Deletion Syndrome (IBBC)
 - 22 sites worldwide participating
 - 12 sites for this study
-
- JAMA Psychiatry, 2015 Apr; 72(4):377-85

•N=829

•IQ (Wechsler)
data

•N=388
 ≥ 2 IQ
measurements,
AND
psychiatric
assessment

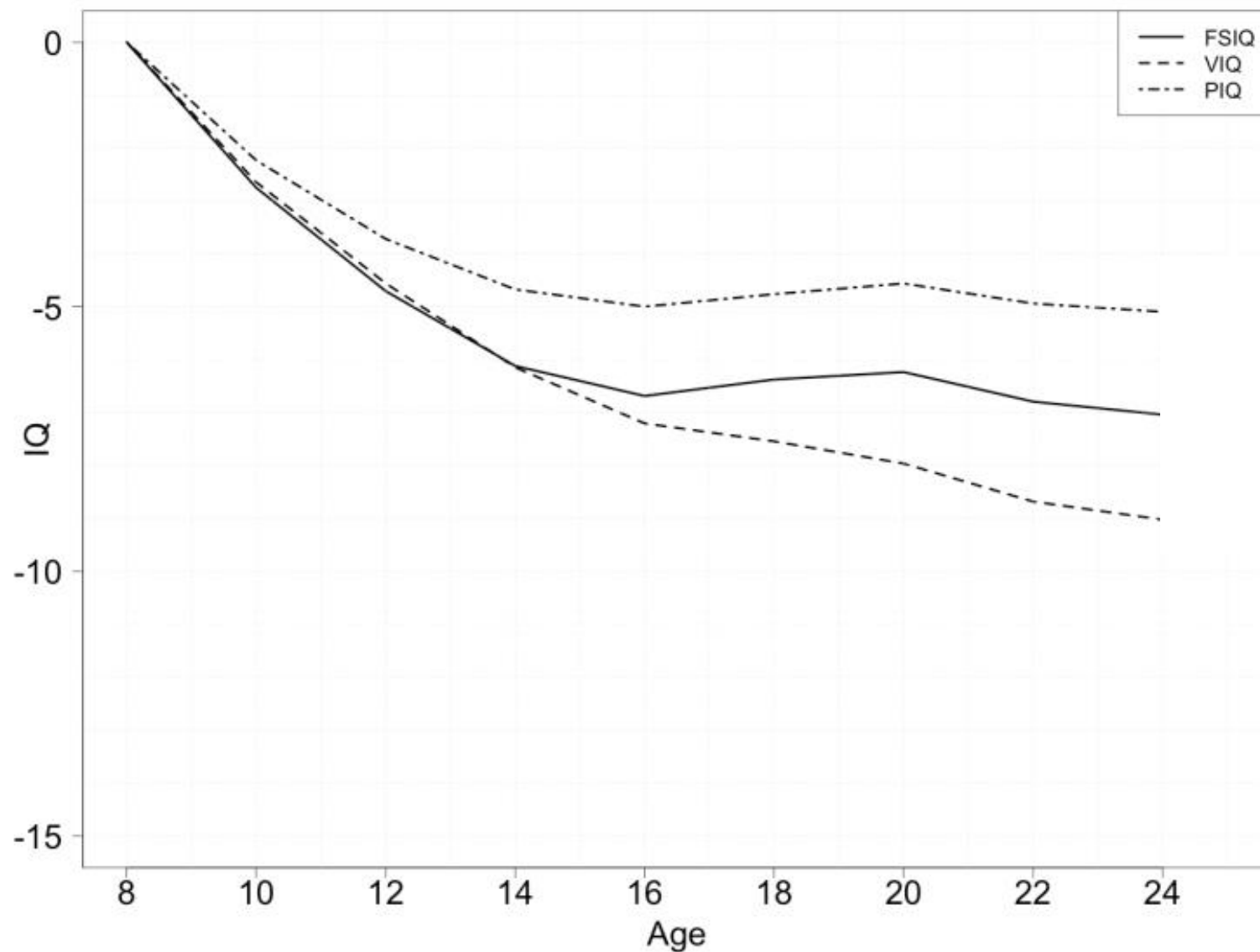
Full scale IQ normative chart for 22q11DS



•Without
psychotic
disorder
(n=341)

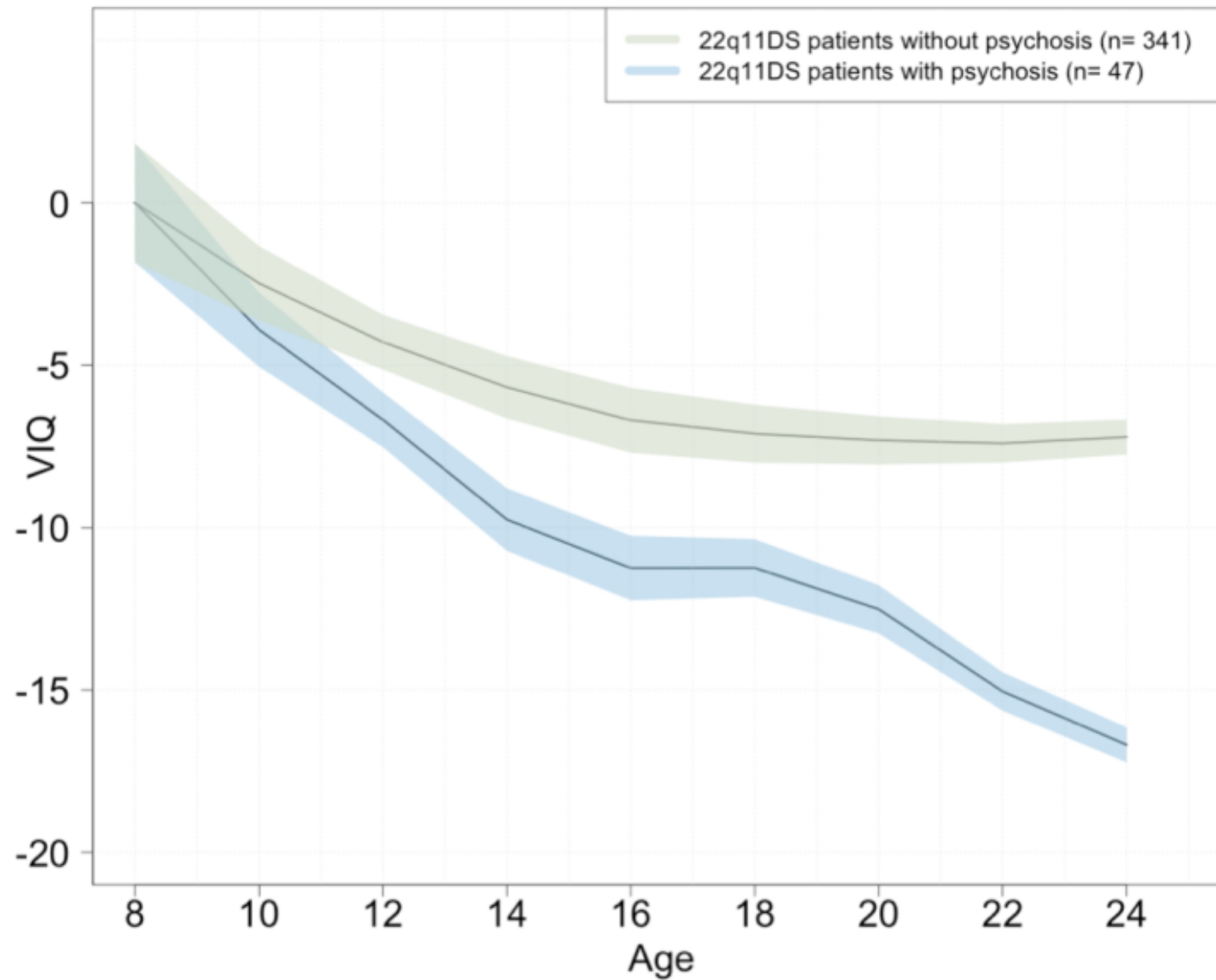
•With
psychotic
disorder
(n=47)

Overall cumulative IQ decline (FSIQ, VIQ, PIQ) in 388 individuals with 22q11DS

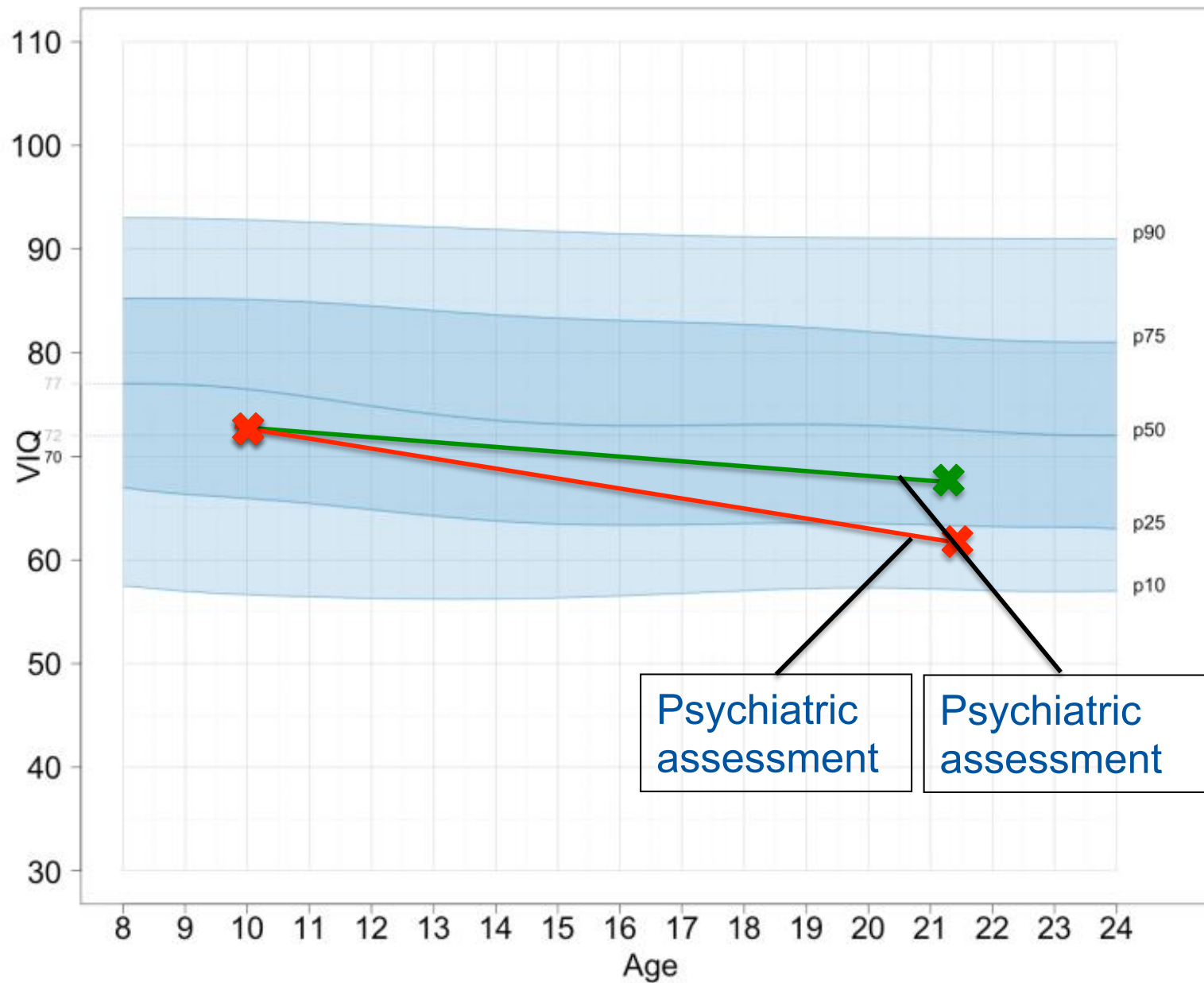


Cumulative decline for verbal IQ

Center



Verbal IQ normative chart for 22q11DS



Our finding is not *novel*



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COMT genotype predicts
longitudinal cognitive decline
and psychosis in 22q11.2
deletion syndrome

Doron Gothelf^{1,2}, Stephan Eliez³, Tracy Thompson¹,
Christine Hinard⁴, Lauren Penniman¹, Carl Feinstein¹,
Hower Kwon⁵, Shuting Jin¹, Booil Jo¹, Stylianos E Antonarakis⁶,
Michael A Morris⁴ & Allan L Reiss¹

NATURE NEUROSCIENCE

Risk Factors and the Evolution of Psychosis in 22q11.2 Deletion Syndrome: A Longitudinal 2-Site Study

Neuroanatomic Predictors to Prodromal Psychosis in Velocardiofacial Syndrome (22q11.2 Deletion Syndrome): A Longitudinal Study

Wendy R. Kates, Kevin M. Antshel, Stephen V. Faraone, Wanda P. Fremont, Anne Marie Higgins,
Robert J. Shprintzen, Jo-Anna Botti, Lauren Kelchner, and Christopher McCarthy

BIOL PSYCHIATRY 2011;69:945–952 947

Conclusions IBBC data



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- The *average* IQ trajectory in 22q11DS is characterized by a modest downward slope.
- In a subgroup: a more prominent decline starts in early childhood (<10 yrs)
- A negative deviation from the (V)IQ 22q11DS normative chart is a risk factor for schizophrenia

Finding & maintaining balance



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Invisible handicap

+

Strong verbal impression (but have poor comprehension)

+

Cognitive decline

-/-

balance



Recommendations



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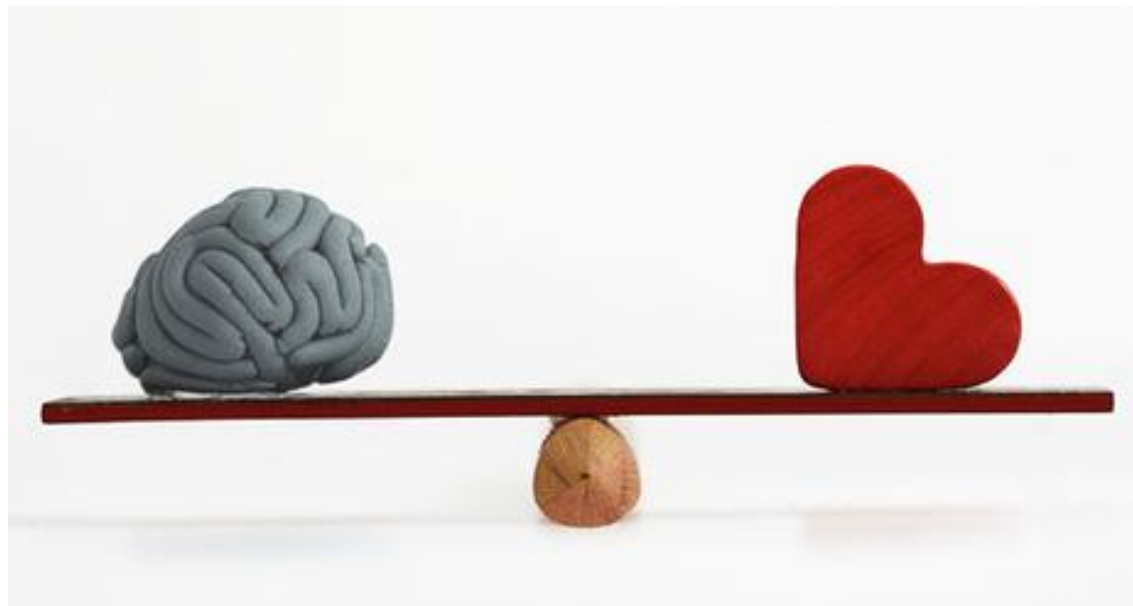
- Regular IQ testing
- Watch out for overdemanding/ overestimation as a result of:
 - Strong verbal impression
 - Strong short term memory
 - Relatively high processing speed
- Regard behavior as a thermometer
 - internalizing behavior
 - school vs home
- Make use of your child's strengths!

Recommendations



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- Your child is unique, follow your heart...
- ...but, make use of the knowledge that is available by regularly consulting the 22q11 professionals. That is what we are here for.



**“Alone we can do so little;
together we can do so much”**



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Sasja Duijff***

& 22q11DS BBC

& Dutch parent association:

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Henriette de Veye
Karin Heestermans
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Sasja Duijff***

Thank you!



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