Research aims
TRIXY National Center of Expertise

Sophie van Rijn, PhD | AXYS 2019 Atlanta, USA
TRIXY Center of Expertise
Trisomy of the X and Y Chromosomes

Clinical Neurodevelopmental Sciences

Treatment and Expertise Center

Academic Medical Center

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TRIXY Expertisecentrum

TRIXY is een nationaal expertisecentrum, waar cliniici en wetenschappers samen werken in de zorg voor kinderen met X en Y chromosoom trisomieën (47,XXY, 47,XXX en 47,XY). TRIXY is een samenwerking tussen de Universiteit Leiden en het Leids Universitair Medisch Centrum.

Meer over het TRIXY Expertisecentrum

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Bringing science to clinical practice and society

Brochures voor kinderen
- Informatie voor meisjes met 47,XXX
- Informatie voor jongens met 47,XXX
- Informatie voor jongens met 47,XY

Brochures voor ouders en volwassenen
- Klinefelter Syndroom, informatie over XXY
- Informatie over Trisomie X (47,XXX)
- Informatie over Klinefelter (47,XY)
- Informatie over 47,XY
- Zorgrichtlijn: Leidraad Klinefeltersyndroom

- Met uw kind de diagnose Trisomie X (47,XXX) bespreken
- Met uw kind de diagnose Klinefelter (47,XY) bespreken
- Met uw kind de diagnose 47,XY bespreken

Brochures voor professionals
- Informatie voor leerkrachten en professionals
- Informatie over Trisomie X (47,XXX)
- Informatie over Klinefelter (47,XY)
- Informatie over 47,XY
- Zorgrichtlijn: Leidraad Klinefeltersyndroom
- Factsheet: Zorg voor kinderen met een extra X of Y chromosoom

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75% of research focused on physical/medical issues (Pieters et al., 2011)
Research cohorts

• 60 adults with XXY
• 60 boys with XXY (6-18 yrs)
• 60 children with XXX or XXY (6-18 yrs)
• 70 (and counting) children with XXX, XXY, XYY (1-6 yrs)
1. Look ‘beyond’ behaviors

• Neurobehavioral symptoms may arise from different underlying cognitive processes
• Why do (a subset of) children with SCT show behavioral symptoms?
• What do we need to monitor/evaluate?
• What are specific targets for support/intervention
A review of neurocognitive functioning and risk for psychopathology in sex chromosome trisomy (47,XXV, 47,XXX, 47, XYY)

Sophie van Rijn* b

Risk for:

- ASD, ADHD, depression, anxiety, bipolar
- Language, executive functioning, social cognition, emotion regulation
XXX, XXY, XYY, aged 1 to 6 yrs

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Behavior

- Questionnaires
- Systematic observations
Top 10 behavior observations of parents

Does your child currently have, or had in the past, any of the following behaviors on a regular basis?

54 %  Tamper tantrums
33 %  Shy
31 %  Short attention span/distractable
27 %  Bothered by things touching him / her
23 %  Immature
21 %  Resistance to change in routines
17 %  Anxiety
15 %  Poor eye contact
14 %  Impulsive
13 %  Moodiness

Important themes: Emotional control, social development, executive functioning

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Cognitive tests
Social perception and executive functioning: Eyetracking
Emotion regulation: Arousal markers in heart rate

- Unexpected events
- In response to others
- When frustrated
- Eye gaze
2. Developmental perspective is key

- Behavioral phenotype is anchored in early neurocognitive development
- Brain continues to develop into 20/30’s
- Cognitive functions become ‘online’
- Growing into deficit?

👉 Key to understand vulnerabilities
👉 Key to optimize outcome
What do we know about early cognitive development in SCT?

- Review study, Clinical Genetics (in press)
- Identifies need for studies focusing on executive functioning, emotion regulation and social cognition
Current research (open for participation)

- 800,000 euro funding in 2016
- XXX, XYY, XXY
- aged 1 to 6 years
- Longitudinal study
- Based at Leiden University
- Collaboration with all academic medical centers in NL and BE
- TRIXY Partner site: XtraordinarY kids clinic, Denver CO

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Prediction over time

Early cognitive functioning  →  Behavioral outcome
International collaboration
eXtraordinarY kids Clinic – TRIXY center
Eyetracking from age 6 months
3. Look ‘beyond’ the extra X/Y chromosome

Genetics of cognitive ability in 11,000 twin pairs (Haworth, 2009):
50% genetic influences
28% shared environment influences
22% unique environment influences

Extra X or Y chromosome
- environmental influences
- especially in childhood
4. Focus also on environmental factors

- Life events
- Socio-economic status
- Parenting styles
- Family functioning
5. Evaluate effect of (preventive) intervention

**Adults** (Martin et al. in revision)
Self-management training focusing on executive functioning in adults with XXY
- improves inhibitory control
- reduces some of the neurobehavioral symptoms

**Children in TRIXY study:**
- Preventive intervention study in The Netherlands
- Early stimulation of socio-emotional development
- Age 3 to 8 yrs
- Home-based DVD training program, daily for 4 weeks
- Pre-post measurements with eyetracking + cognitive tests
We thank all participating families!

Interested in participating?
TRIXY Denver site
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