

Quality of Life in Children, Adolescents and Adults with Cystinosis

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1. Quality of Life (QoL) and cystinosis
2. Results of multicentric study
3. Psychological interventions to enhance QoL



QoL and Cystinosis



- Definition QoL
- What does literature say?
 - Results of studies in Chronic Kidney Disease (CKD)
 - Results of studies in Cystinosis
- Clinical impression: Cystinosis and QoL through development



- Definition QoL:

*HRQOL is a **multidimensional construct** that consists of at least **three broad domains** – physical, psychological, and social functioning – that are affected by one's disease and/or treatment. **Physical functioning** is usually defined as the ability to perform a range of activities of daily living, as well as physical symptoms resulting from the disease itself or from treatment. **Psychological functioning** ranges from severe psychological distress to a positive sense of well-being and may also encompass cognitive functioning. **Social functioning** refers to quantitative and qualitative aspects of social relationships and interactions and societal integration.*

Literature: QoL in Chronic kidney disease?

- Next to medical treatment, QoL is perceived as one of the most important goals of patient care
- QoL in children with CKD < QoL healthy children
- Child report > Parent-report

Intellectual and motor performance, quality of life and psychosocial adjustment in children with cystinosis

**Francis F. Ulmer • Markus A. Landolt •
Russia Ha Vinh • Thierry A. G. M. Huisman •
Thomas J. Neuhaus • Bea Latal • Guido F. Laube**

Mood and illness experiences of adults with cystinosis

**Naomi Beinart, Ruth A. Hackett, Christopher D. Graham, John Weinman &
Marlies Ostermann**

Quality of Life is Improved and Kidney Function Is Patients with Nephropathic Cystinosis Treated for 2 Years with Delayed-Release Cysteamine Bitartrate

**Craig B. Langman, MD¹, Larry A. Greenbaum, MD, PhD², Paul Grimm, MD, PhD³, Minnie
Sarwal, MD, PhD³, Patrick Niaudet, MD⁴, Georges Deschenes, MD, PhD⁵, Elisabeth A. M.
Cornelissen, MD⁶, Denis Morin, MD⁷, Pierre Cochat, MD⁸, Ewa Elenberg, MD, MS⁹,
Christian Hanna, MD, MS¹, Segolene Gaillard, MD¹⁰, Mary Jo Bagger, MS¹¹, and Patrice
Rioux, MD, PhD¹¹**

Study 1:

**Intellectual and motor performance, quality of life
and psychosocial adjustment in children with cystinosis**

- 11 patients with cystinosis
- Mean age: 9,7 years (5,3 – 19,9 y)
- Results:
 - Patient- report: +- normal QoL
 - Parent-report: lower QoL in positive emotions, autonomy, social and cognitive functioning
- Children rated their own QoL more optimistically than their parents!

Study 2:

Mood and illness experiences of adults with cystinosis

- 23 patients with cystinosis (18-47 years, 11 males)
- Questionnaire T1: Hospital Anxiety and Depression Scale
- Interview T2 (12 months later)
- Results questionnaire:
 - Mean level of anxiety and depression higher than other patient groups (e.g., general medicine), but below psychiatric outpatients

Study 2:

Mood and illness experiences of adults with cystinosis

- Results Interview: Three themes
 - Theme 1: physical impact
 - Theme 2: impact on relationships, autonomy and social life
 - Theme 3: coping strategies

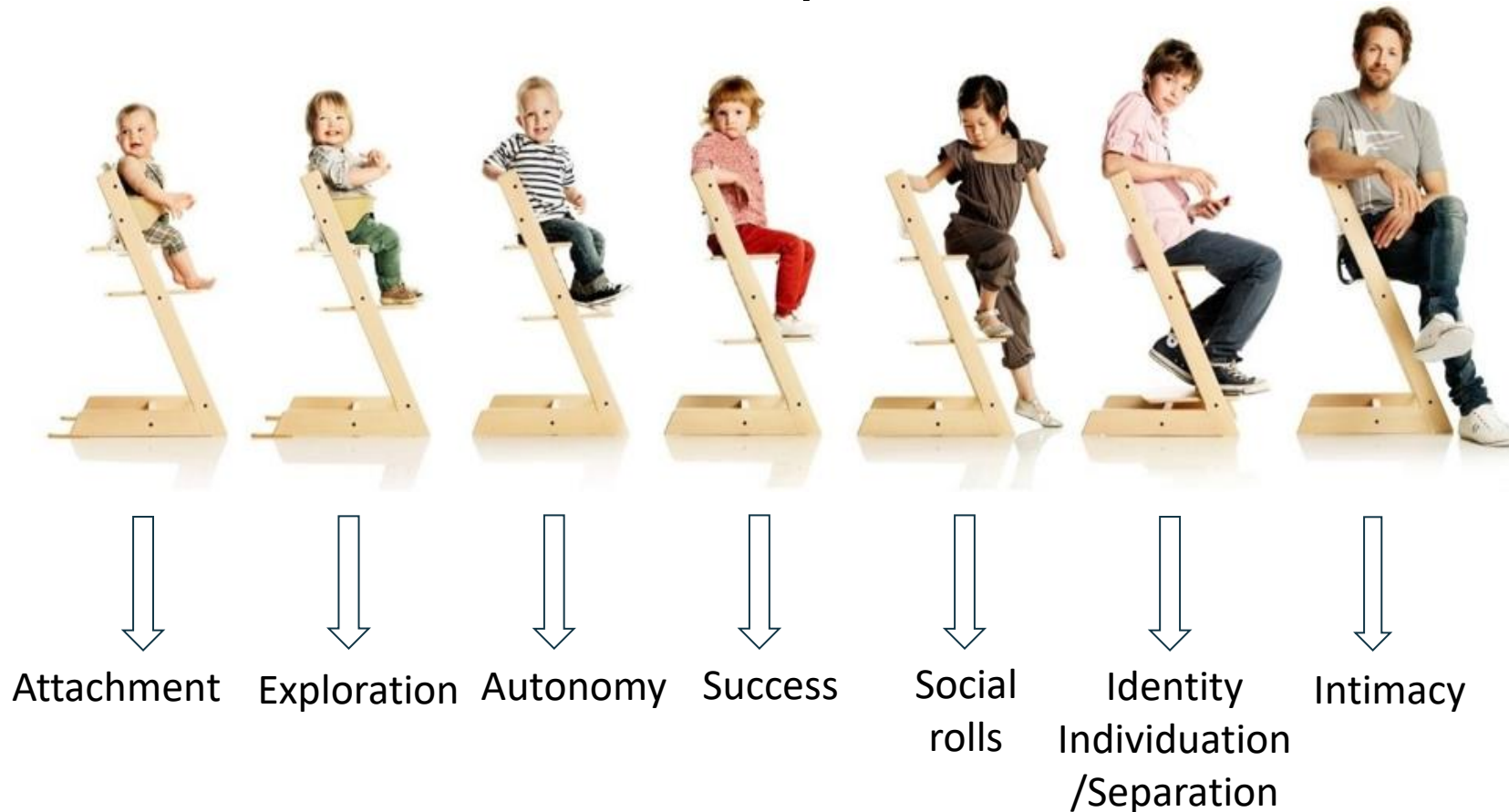
→ Although there were many difficult aspects, many positive elements were reported by participants!

Study 3:

Quality of Life is Improved and Kidney Function Preserved in Patients with Nephropathic Cystinosis Treated for 2 Years with Delayed-Release Cysteamine Bitartrate

- Aim: determine long-term effects of delayed-release cysteamine bitartrate (every 12 hours) compared with immediate-release cysteamine (every 6 hours)
- 40 patients, average age 12 years
- Results:
 - Significant improvement in social function, school function, and total function after delayed-release cysteamine

Chronic illness and child development



- Infancy - toddlerhood:
 - Uncertainty about diagnosis
 - Feeding problems of the young child
 - Frequent hospitalization: impact on development, stress in the parent
 - Struggle about medication intake
 - Anxieties
- Latency:
 - School performance problems
 - Feeling different than peers



Examples of clinical impression: QoL in cystinosis

- Adolescence:
 - Illness identity
 - Social struggles
 - Adherence problems
 - Individuation/separation
 - Transition
- Young adulthood:
 - Autonomy, independence,
 - Intimate relationships
 - Studies, work,..
 - Transition



But also: Tremendous resilience:

- Brave!
- High capacity to respond positively to the adverse situations!
- Optimism and positive mood!
- ...

Examples of clinical impression: QoL in cystinosis

What about the parents....

REVIEW ARTICLE

Experiences of Parents Who Have Children With Chronic Kidney Disease: A Systematic Review of Qualitative Studies

Allison Tong, MPH^{a,b}, Allison Lowe, BSc^a, Peter Sainsbury, PhD^{b,c}, Jonathan C. Craig, PhD^{a,b}

Parental perspectives on caring for a child with chronic kidney disease: an in-depth interview study

A. Tong,^{*†} A. Lowe,^{*} P. Sainsbury^{†‡} and J. C. Craig^{*†§}

^{*}Centre for Kidney Research, The Children's Hospital at Westmead, Westmead

[†]School of Public Health, University of Sydney

[‡]Population Health, Sydney South West Area Health Service, Sydney, and

[§]Department of Nephrology, The Children's Hospital at Westmead, Westmead, NSW, Australia

- 4 main theme's:
 - Absorbing clinical environment
 - Medicalising parenting
 - Coping strategies and support system
 - Disrupting family norms

Study Family adjustment in children with cystinosis

BEHAVIOR AND ADJUSTMENT IN CYSTINOSIS

75

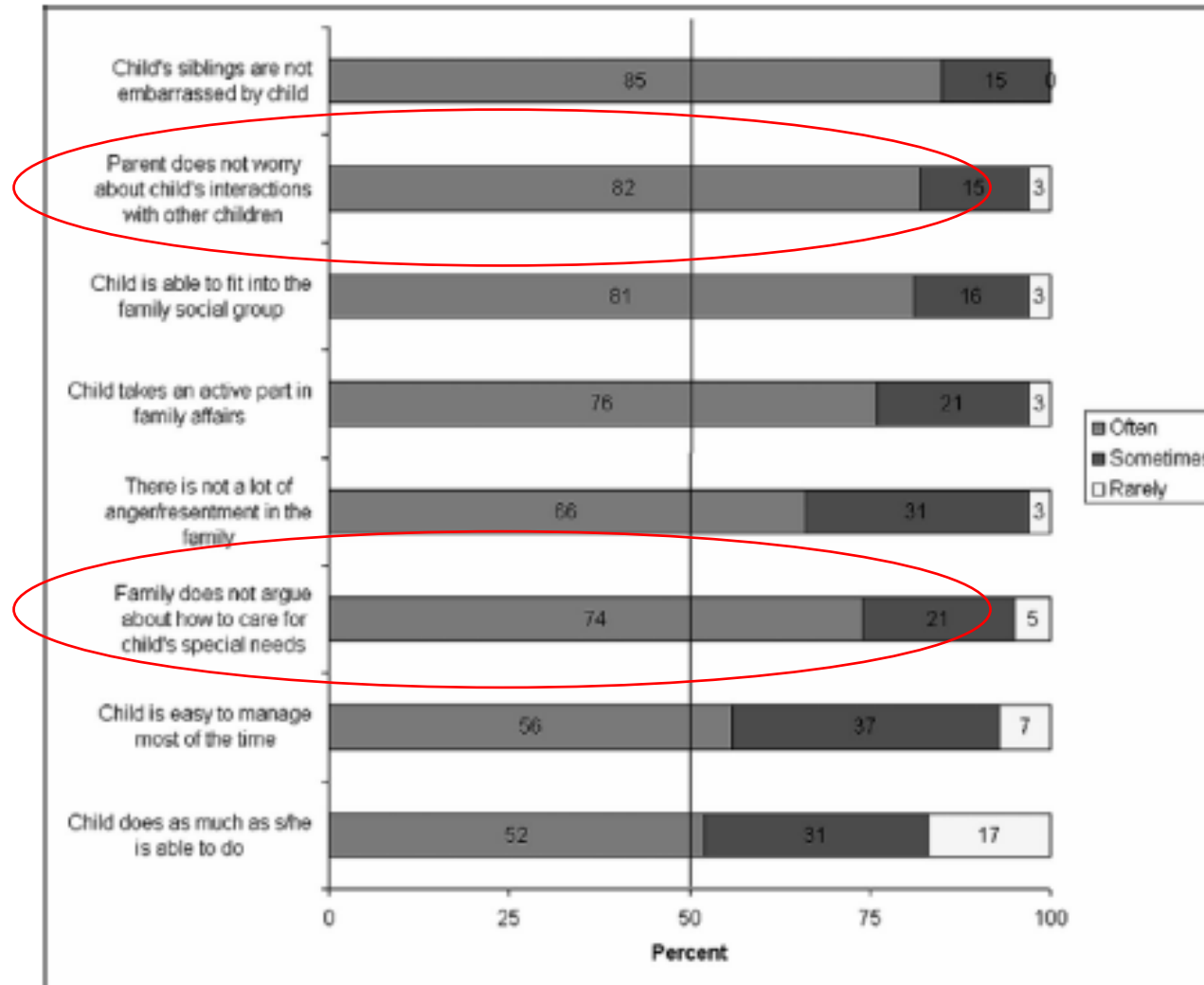


Figure 3. Percentage of Parents Responding "Often or Very True," "Sometimes or Somewhat True," or "Rarely or Not True" to Family Positives/Strengths on the Cystinosis Behavior Questionnaire.

Study Family adjustment in children with cystinosis

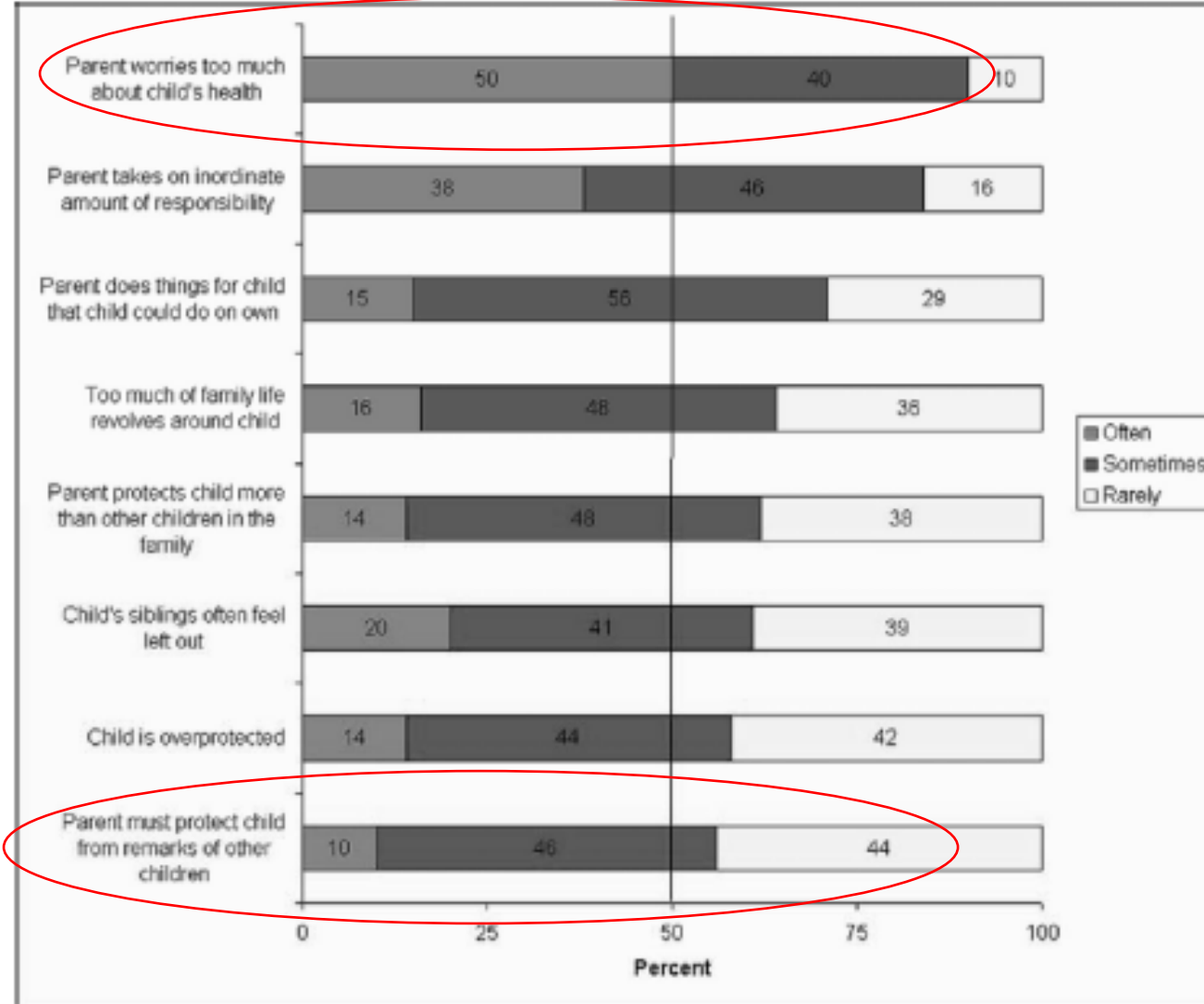


Figure 4. Percentage of Parents Responding "Often or Very True," "Sometimes or Somewhat True," or "Rarely or Not True" to Family Negatives/Stressors on the Cystinosis Behavior Questionnaire.

Spilkin & Ballantyne, 2007

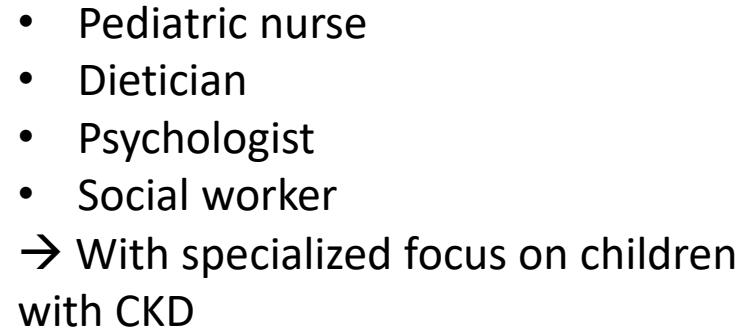
Results of the Study:

A Multicentric Belgian Study on Illness-related Parental Stress and Quality of Life in Children with Severe Kidney Diseases

With funding of Fonds Kind en Orgaantransplantatie Prof. Dr. R. Van Damme-Lombaerts



- UZ Leuven
- UZ Gent
- UZ Antwerpen
- UKZKF/Huderf
- UCL Saint Luc
- CHC de Liège



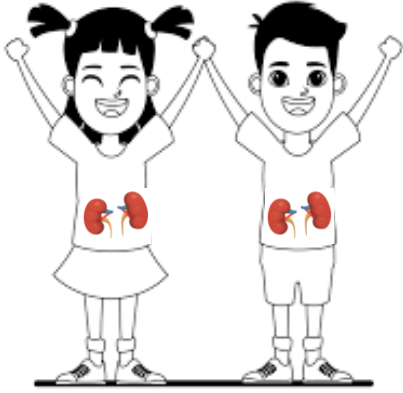
Data collection in every center to investigate:

- QoL of the child
- Illness-related stress in parents of children with CKD



Aim of the present study:

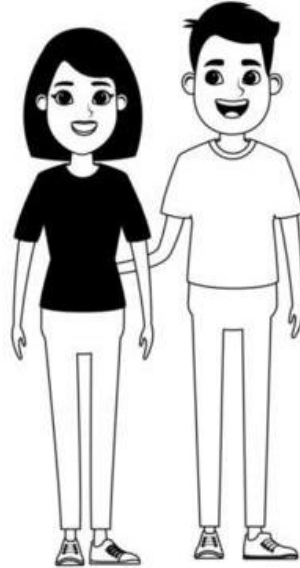
- Compare the levels of child's QoL and illness-related parental stress between different kidney disease categories
- Explore the associations between illness-related parental stress and child quality of life



$N = 295$
176 boys, 119 girls
 M age = 11.8, $SD = 3.7$
Range = 0 – 18 years

Kidney Disease Categories:

- Structural kidney diseases = 42 (14%)
- Tubulopathies and metabolic diseases = 53 (18%)
- Nephrotic syndromes = 69 (23%)
- Acquired diseases with proteinuria and hypertension = 74 (25%)
- Kidney Transplantation = 57 (19%)



$N = 285$

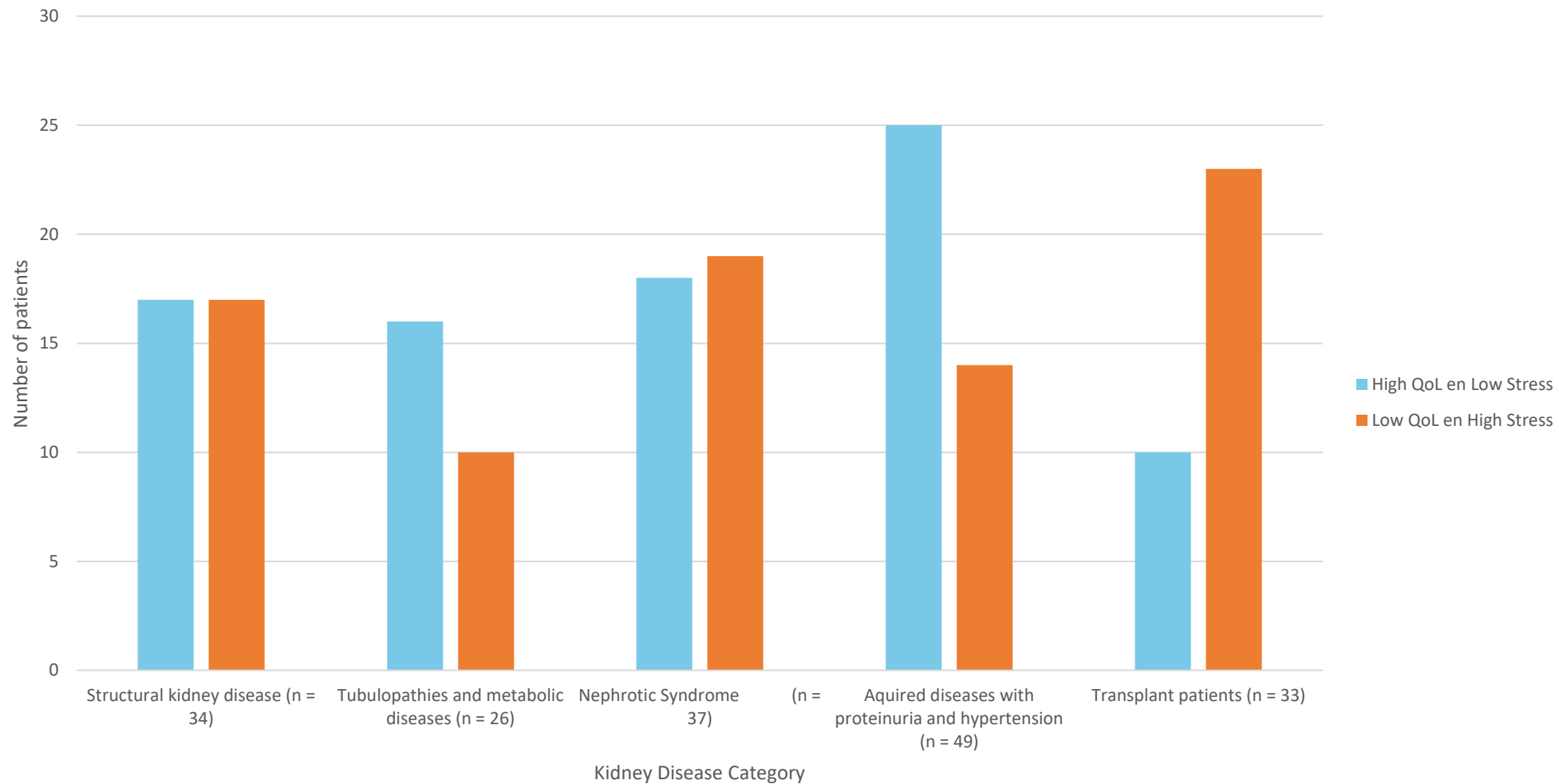


Questionnaires

- Illness-related parental stress: Pediatric Inventory for Parents (Vrijmoet-Wiersma et al., 2010)
- Quality of Life: PedsQL™ Core 4.0 (Varni, 1998)

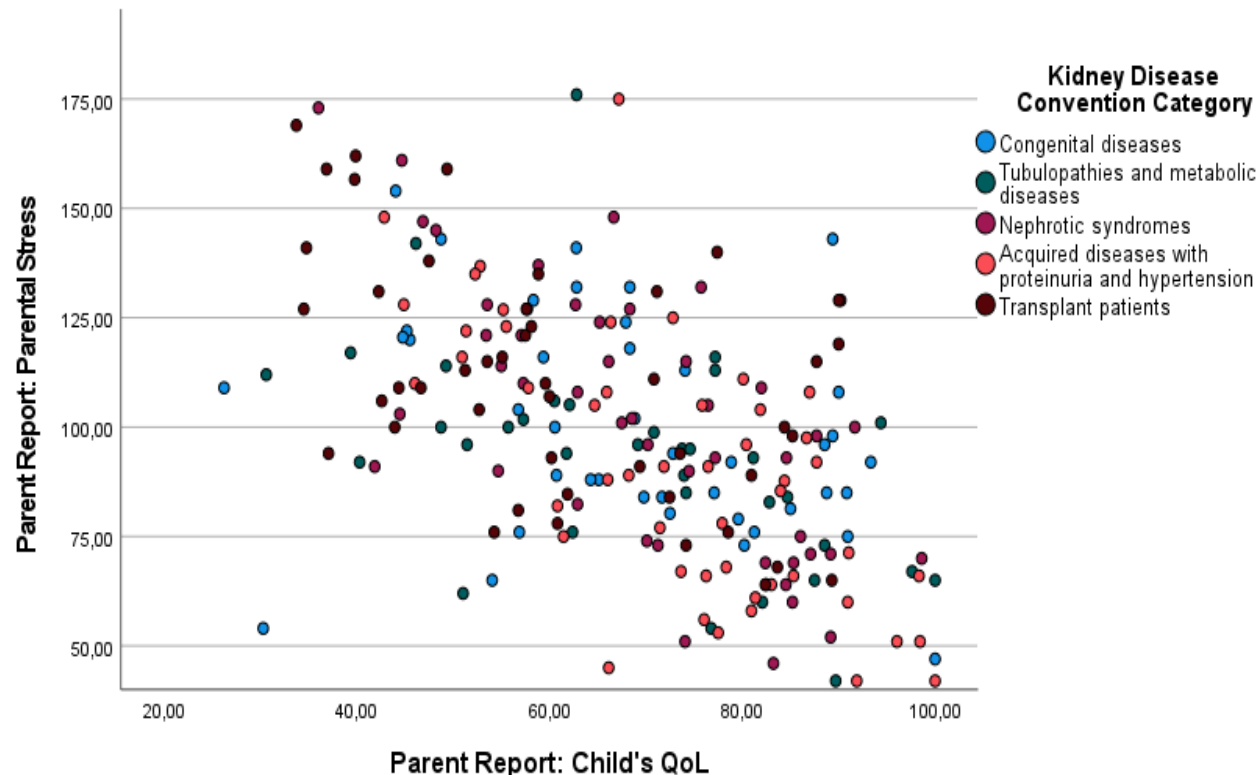
1. Comparison of child QoL and parental stress between different kidney disease categories

- Child report: no differences in QoL between kidney diseases ($p > .05$)
- Parent report: sign differences in QoL between kidney diseases ($p = .02$)
- Sign differences in parental stress between kidney diseases ($p = .02$)
- Parents of transplanted patients report lower child QoL and higher parental stress



2. Associations between illness-related parental stress and child quality of life

- Negative associations between parental stress and child QoL





Summary of results:

1. Significant differences in QoL & parental stress between kidney disease categories, when based on parent reports
2. Children rated their QoL more optimistically than their parents
3. According to parents (not to the child): Lowest child QoL and highest parental stress in transplant group
4. More parental stress is associated with worse QoL in the child

Future research

1. Develop cystinosis specific QoL
2. Focus in detail on cystinosis patients

These results highlight the importance of multidisciplinary care for children with severe kidney diseases

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Psychological interventions: How can we help?



- Help with treatment/medication regimen:
 - Education about disease, help with practical aspects of therapy in daily life, talk about side effects of cysteamine and find solutions,..
 - Facilitators, organizational strategies (e.g., simplifying regimens, reminders,..) → MyNexuz, Cystinosis & Me App!



- Psychological interventions:
 - Screening and follow-up
 - Help with coping strategies, provide psychological support
 - Help with compliance difficulties
 - Encourage to talk to others about the disease, ask for help, talk to friends, parents, team...
 - Transition interventions
 - Support for the parents!!!



- Interventions at school:
 - Provide information and education about cystinosis and the impact of the disease to children and teachers (with permission!)
 - **New!!! My Cystinosis Story: Presentation toolkit for children with cystinosis**
 - Help with special arrangements (more time, TOAH,...)
- Social support:
 - Help adolescent to talk to friends, ask for support
 - Support of the team
 - Support groups for both children and parents



- Cystinosis has an impact on QoL
- Many studies indicate that children rate their QoL more optimistically than their parents
- Studies also indicate positive aspects!
- Importance of a multidisciplinary team to support patients and families with cystinosis!



My Cystinosis Story

Presentation toolkit for children with cystinosis

A collaboration between:

- **UZ Leuven, UZ Gent, UCL, CHU Liege, UZA Antwerpen**

Prof. Raes, Prof. Rangelov, Ari Van Hulle, Lore Willem,
Dr. Collard, Dr. Veys, Prof. Levtchenko, Prof. Trouet

- **Patients and patient association**

Visiting patients and their parents, Marjolein Bos, Fons Sondag

- **Service design agency**

Ideate



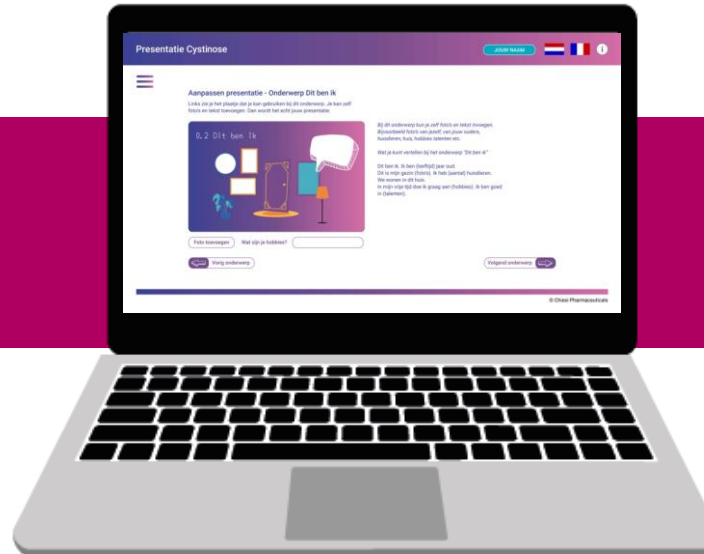
My Cystinosis Story

Presentation toolkit for children with cystinosis

My cystinosis story contains:



Doll for demonstration of the organs



Website with slide builder containing images and text to create your personal story about the disease



Sessions with presentation coach Veronique Hoex for confidence and practice

My Cystinosis Story

Presentation toolkit for children with cystinosis

How it works:



At the hospital a nurse hands you a doll



At home you read the information on the back of the doll's container



You visit the website and build your own story



You practice your story with a coach and become a star at presenting!

Now you are able to share with anyone who you are, what you are going through and how others can help!

My Cystinosis Story

Presentation support for children with cystinosis



Introduction of the Coach!



Presentation of Marte!

<https://photos.app.goo.gl/vBxnt2ghkraQMA5h8>

Thank you for your attention!



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