HoFH WEBINAR TRILOGY

Advancing Knowledge, Care, and Community

Living with HoFH Reality: Through the Lens of Children, Parents and Siblings

Matthew Bolz-Johnson, Chiesi UK & Ireland

Chiesi UK has been invited to speak at the webinar and funds the speaker to present. The content of the presentation is based on previous published evidence of the speaker.

FH Europe Foundation

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Disclosure

Matt Bolz-Johnson, Patient Advocacy & Public Affairs Lead, Rare Diseases Team

I am a full-time employee of Chiesi UK & Ireland, and I have been invited to speak today by FH Europe.

What is Mental Health?

Mental health is a basic human right. Mental health is defined as:

“A state of mental well-being that enables people to cope with the stresses of life, realize their abilities, learn well and work well, and contribute to their community” (WHO)

It is an **integral component of health and well-being** that underpins our individual and collective abilities to make decisions, build relationships and shape the world we live in. And it is crucial to personal, community and socio-economic development.

Antonietta, Italy

Reference: World Health Organization (2025) Mental health: strengthening our response. Available at: WHO fact sheet

Psychosocial Risk & Protection Factors

Reference: Bolz-Johnson et al. 2026

Daily Life & Constant Adaption

Diagnosis Odyssey

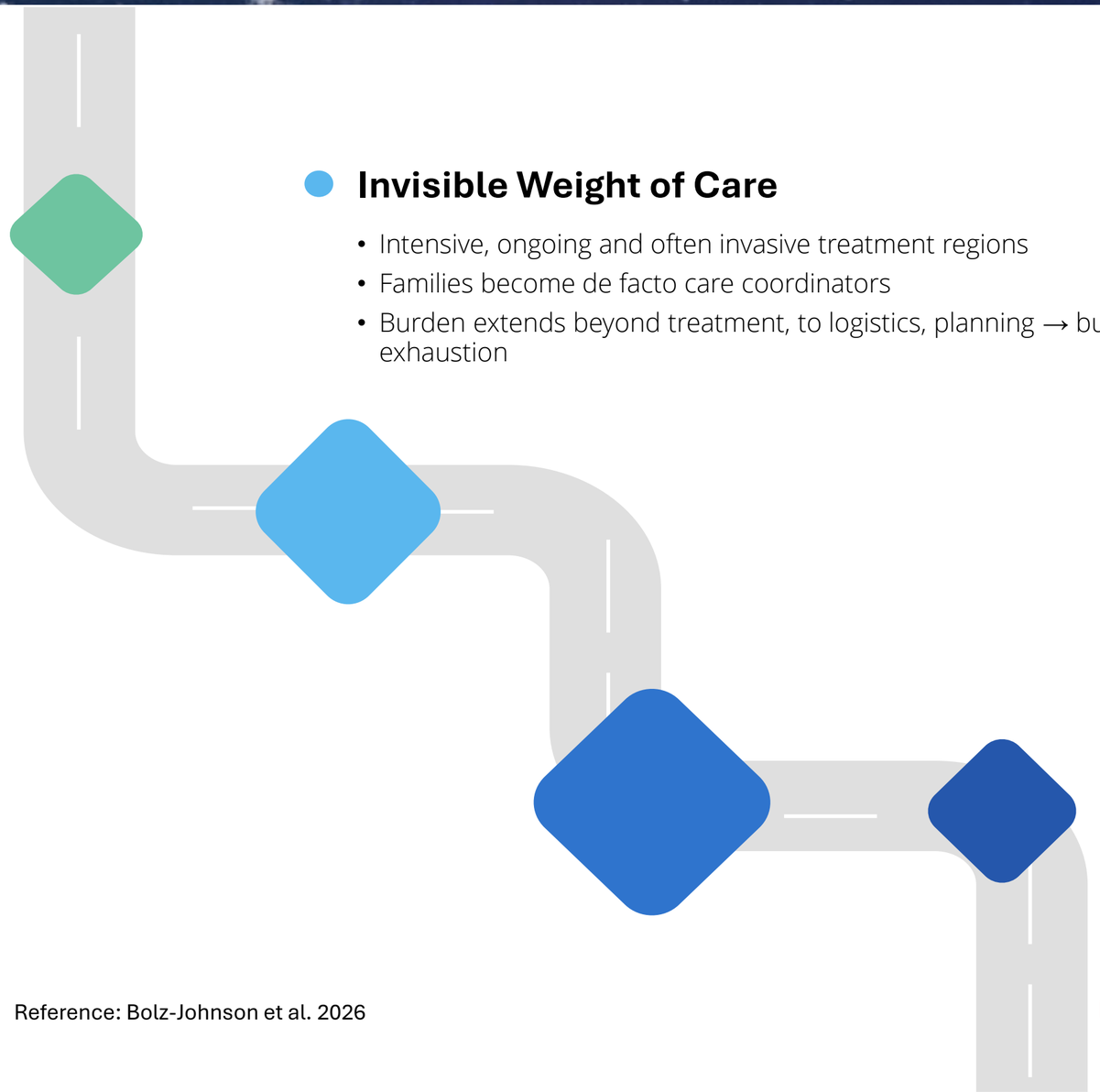
- Lengthy diagnosis, paved with history of misdiagnosis
- Poor communication impacts trust in HCP's
- Fragmented, uncoordinated care resulting in high logistical burden

Living with multiple uncertainty

- Daily life is dominated by self-management and unpredictability.
- Ongoing uncertainty about prognosis and treatment.
- Low awareness increases isolation and frustration.

Stress & Strain on Life & Relationships

- Lack of understanding from wider social networks
- Social isolation and reduced participation
- Strain on relationships and family functioning
- Increased exposure to stigma and discrimination



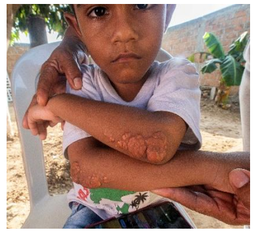
Invisible Weight of Care

- Intensive, ongoing and often invasive treatment regions
- Families become de facto care coordinators
- Burden extends beyond treatment, to logistics, planning → burnout, exhaustion

Reference: Bolz-Johnson et al. 2026

Unmet Needs

Psychological distress is a *typical response to the reality of living with a rare condition*, not pathology, (Bolz-Johnson et al. 2026)



Emotional Wellbeing

~70% of PLWRD reported poor mental health.

~30% had suicidal thoughts in past 6 months.

(Rare Barometer Survey Results 2026)

Social Isolation

54% people living with a rare disease declare that the rare disease caused or amplified isolation from friends and family.

(Courbier et al. 2017)

Financial Independence

7/10 PLWRD have reduced or stopped professional activity due to their own or their family members' disease.

(Courbier et al. 2017)

Psychological Support

75% received no professional psychological support at diagnosis.

(Rare Barometer Survey Results 2026)

Note: People Living with a Rare Disease (PLWRD)

Marlon, Colombia; Antonietta, Italy, and; Max & Moi, Colombia

Family as the Care System

Rare conditions affect the entire family unit, not just the individual

Family functioning is a key determinant of quality of life and mental health outcomes

(Boettcher J et al 2020)

Maria Grazio, Italy

Caregivers experience:

- Emotional distress, fatigue, and burnout
- Financial and employment challenges
- Limited access to support services

Parents often:

- Prioritise the child over their own wellbeing
- Require support for uncertainty, decision-making, and care coordination

Reference: Bolz-Johnson et al. 2026; Bolz-Johnson et al 2026.

Siblings: The Invisible Experience

Siblings experience psychological and emotional challenges that are often unrecognised

Common sibling experiences:

- Feeling overlooked or “invisible”
- Balancing caregiving roles with normal childhood
- Anxiety about the future
- Managing complex emotions (guilt, worry, isolation)

Children and adolescents with rare conditions:

- Struggle with identity and feeling “different”
- Transition often involves redefining a stigmatised identity while learning to navigate independence, managing new autonomy and the responsibility of facing an uncertain future.

→ Need for tailored psychological and social support for siblings

Antonietta, Italy

Reference: M. Bolz-Johnson 2026

Gendered Impact: Women, Care and Inequity

Maria Grazia, Italy

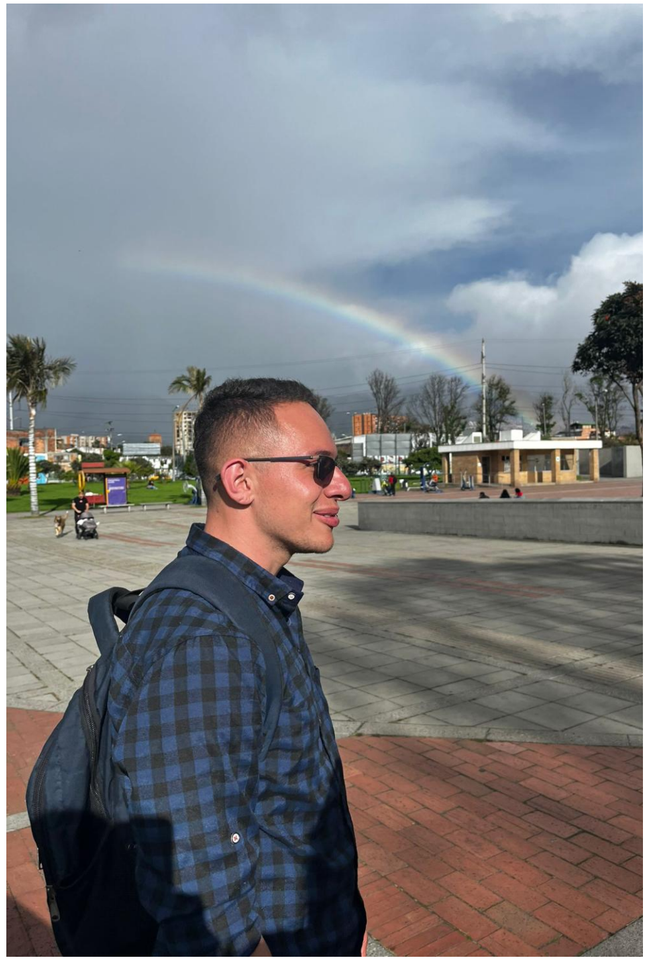
- Women are often primary caregivers, bearing disproportionate psychological and practical burden.
- Gender roles influence access to support, wellbeing, and coping.

- Women more likely to:
  - Carry caregiving responsibilities alongside other roles
  - Experience cumulative stress and reduced wellbeing

- Broader inequities include:
  - Cultural barriers and stigma affecting access to care
  - Under-recognition of caregiver mental health needs

(Bolz-Johnson et al. 2026)

Key Take Away Messages



Marlon, Colombia

- Living the HoFH reality means navigating a lifelong, family-wide impact, medical, psychological, and social, that remains insufficiently recognised in care systems.

- HoFH is not just a cardiovascular condition, it is a lifelong psychological journey affecting identity, wellbeing, and coping
- Treatment burden in HoFH is cumulative and systemic, not just clinical
- HoFH is experienced every day, not just in clinics
- Managing uncertainty requires continuous adaptation and coping strategies
- The family is the primary care infrastructure in rare disease
- Rare disease creates a shared but unequal burden across siblings
- Gender dynamics shape the experience of HoFH within families

Reference: Bolz-Johnson et al. 2026.

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Thank You

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