The Integration of Family System Based Research Programmes in Genetic Rare Diseases

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Rare Diseases 360
Collaborative Strategies to leave no-one behind
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Dr Suja Somanadhan: Assistant Professor of Children's Nursing, UCD, Dublin, Ireland, Prof Thilo Kroll: Professor of Health Systems Management, UCD, Dublin, Ireland, Prof Philip Larkin: Professor of Clinical Nursing (Palliative Care), UCD, Dublin, Ireland

Background

- When a child is diagnosed with a rare genetic disease, the overall responsibility for the ongoing complex care and management relies on the family, particularly on the child's parents.
- Parenting can have a significant impact on parents' lives, creating physical and emotional challenges for them, siblings and other close family members including grandparents.
- Existing studies highlight that many families have unmet needs of services and care during their child's lifespan of living with rare diseases and beyond and many find it very difficult to navigate the health system to identify what help is available and from where is a huge challenge for the families of children with rare diseases.

Key Features

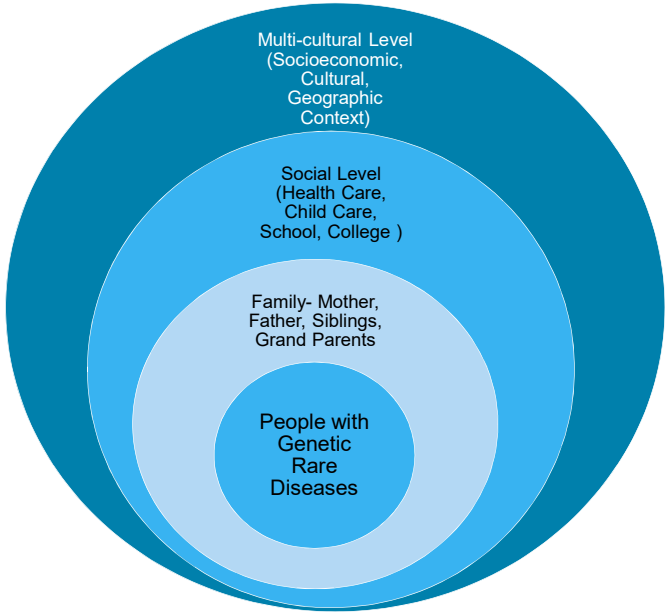
- Symptoms interfere with many normal activities & routines
- Medical treatment is restricted in it's effectiveness
- Treatment itself contributes to the disruption of activity of daily living
- These children and their families have immense needs
- Most of these children live at home with their families

Overview

This poster will highlight the integration of family system based research programmes for the children, young people and their families and healthcare professionals experience of living with and caring for people with rare genetic diseases.

Family Systems Theory

Bronfenbrenner's Ecological Systems Theory Model



Living with Rare Diseases

(Somanadhan & Larkin, 2016)

Research Programmes

Research 1

Families experience of living with rare diseases (2010-2016)

Research 2

Measuring the impact of caring for children with rare diseases (Parents /HCPs)(2017-Present)

Research 3

Measuring the social needs of families of children with rare diseases (2018-Present)

Research 4

To understand children and young people experience of living with rare diseases (2017-Present)

Conclusion

- This innovative family system research proposes to offer a better understanding of the experiences encountered by children and their Parents' Day-to-day challenges of living with a rare condition, which will, in turn, guide health service providers in developing care pathways and support services for the patients and their families with rare diseases.
- Provides evidence for policy makers to improve the service delivery for the children with MPS and other rare diseases and their families.
- It will increase healthcare professionals' knowledge of the-day-to-day experiences of living with rare diseases.
- It will support, communicate with and advise patient rare-disease networks and organisations on unmet parent needs, to shape family-centered care practices.

Acknowledgement

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- Should you have any queries or require any further information please do not hesitate to contact us: email: suja.somanadhan@ucd.ie follow on Twitter @sujas15