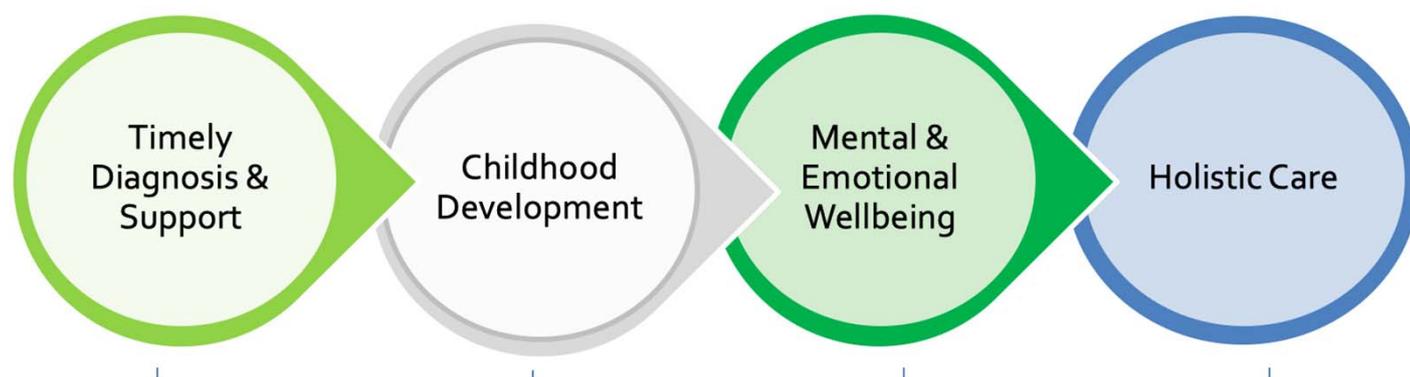


Patient Journey Common Needs: Rare congenital malformations + syndromes with intellectual and other neurodevelopment disorders

INTRODUCTION

Rare congenital malformations and syndromes with intellectual and other neurodevelopment disorders mainly affects one child in a million. The heterogeneous nature of their clinical presentation and huge variation in first symptoms that can present from the newborn period, often result in a delay in securing a timely diagnosis as well as without any available treatment options. Many are common symptoms seen across different syndromes, some are more syndrome-specific. ERN ITHACA has been set up to improve the prognosis and quality of lives for the children affected. But with several thousand rare diseases, how can the Network response to the wide and diverse needs of these children? The patient representatives (ePAG) mapped out the individual needs of their children in a syndrome specific 'patient journey' that acted as a personal testimony and mapped the natural history of each of the rare syndromes.



METHODOLOGY

- Patient representatives presented their syndrome-specific 'patient journeys' (Bolz-Johnson et al. 2019) in a workshop and identified the characteristics and needs that were common.
- The common needs were developed into a position paper that was relevant for all syndromes and summarized under key headings.
- The common needs were presented to ERN ITHACA at the Network's Annual Meeting in Düsseldorf, 2019.

RESULTS

Genetics is the key for many ITHACA syndromes however it is only one aspect of child and ERN ITHACA needs to go beyond the genetic diagnosis and think about the holistic needs of this patient community. Key needs include:

Timely Diagnosis & Support:

- Early diagnosis and family-centred communication and support of ITHACA syndrome
- The corner-stone to optimise family's ability to best cope and independence.

Childhood Development:

- optimizing childhood development through access to therapies
- enable personal assistance and equipment to ensure mobility and independence (self-care) living

Mental & Emotional Wellbeing

- Mental and emotional wellbeing
- Psychological support for the children AND their parents.
- Psychiatric support is vital for many syndromes in early childhood and in adulthood.

Holistic Care

- Integrated care planning, case management and care coordination across the life-journey of our children
- Social care and educational support
- Ensuring continuity of care through transition to adulthood
- Annual MDT one-stop-shop health check

CONCLUSIONS

Recommendations based on patients' experience have been developed for care & therapies, treatment, psychological support, transition of care, professional training and education. An agreement with experts on introducing common needs in all the workshop of ERN ITHACA have been achieved at the board meeting in Dusseldorf, as well as developing a standards of care in a Clinical Pathway for children with a rare disease. –