



Assessment of Nutrition Care Needs in the Management of Pediatric Patients with Rare Disease

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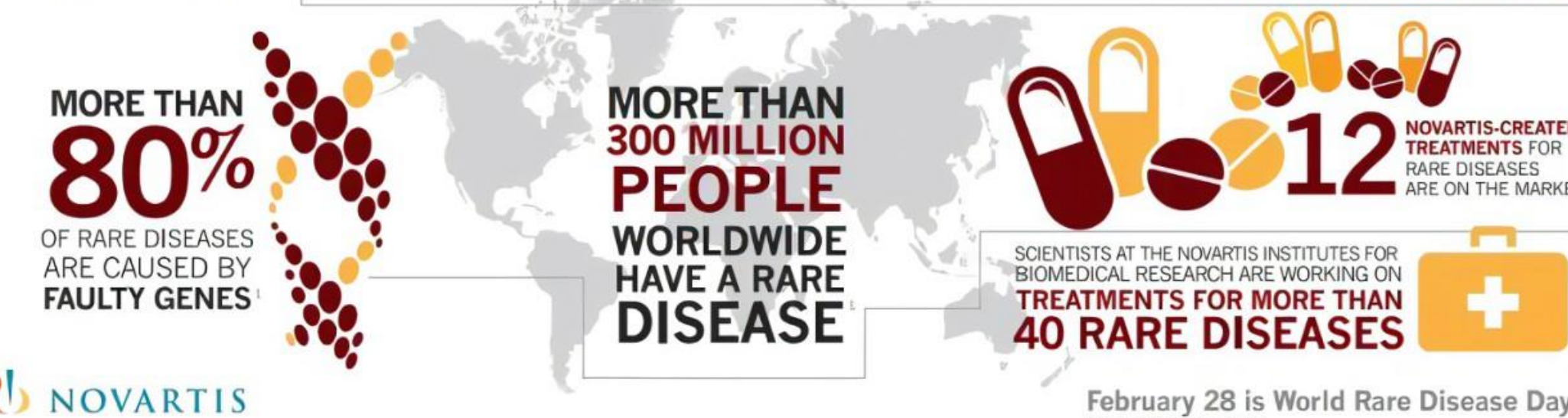
ABSTRACT

Over 1,200 diseases are listed in the National Organization for Rare Disorders database. According to the Government Accountability Office, ~30 million people in the U.S. –half of them children– have rare diseases. Many of the diseases affecting pediatric patients can be attributed to inborn errors of metabolism. The biomechanical pathways underlying these errors typically involve genetic aberrations that cause deficiency of key enzymes responsible for maintaining metabolic homeostasis. Consequently, patients diagnosed with such syndromes can have increased and/or unique nutritional needs, reduced quality of life, and poor prognoses. Our research seeks to formulate best practices for nutritional intake in patients with rare disease. Case studies demonstrate complications and barriers in obtaining nutritional requirements for these patients, possibly due to the limited availability of existing research for practitioners to reference. Additional challenges include the daily limitations of a caregiver to a rare disease patient. Because of the many variables involved in treating these patients, definitive guidelines do not exist thus far.

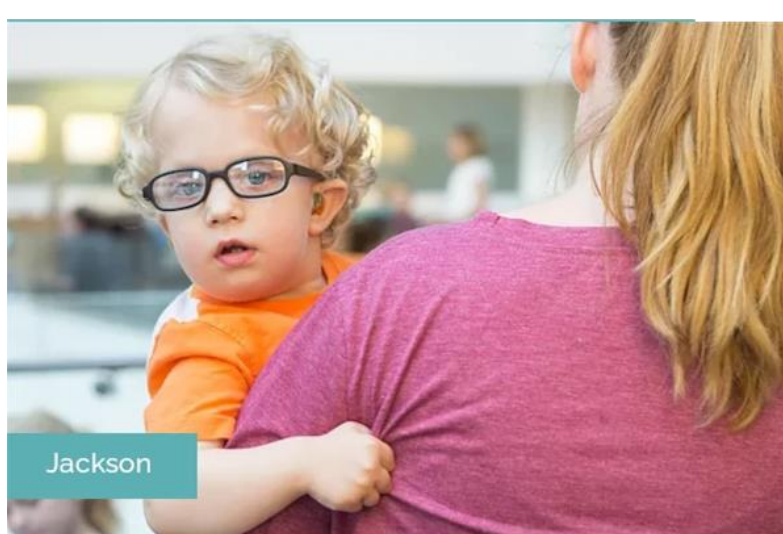
PURPOSE

RARE DISEASES: MORE COMMON THAN YOU THINK?

Rare diseases are defined as those affecting a small percentage of a population – fewer than 200,000 in the U.S. and fewer than 1 in 2,000 in Europe



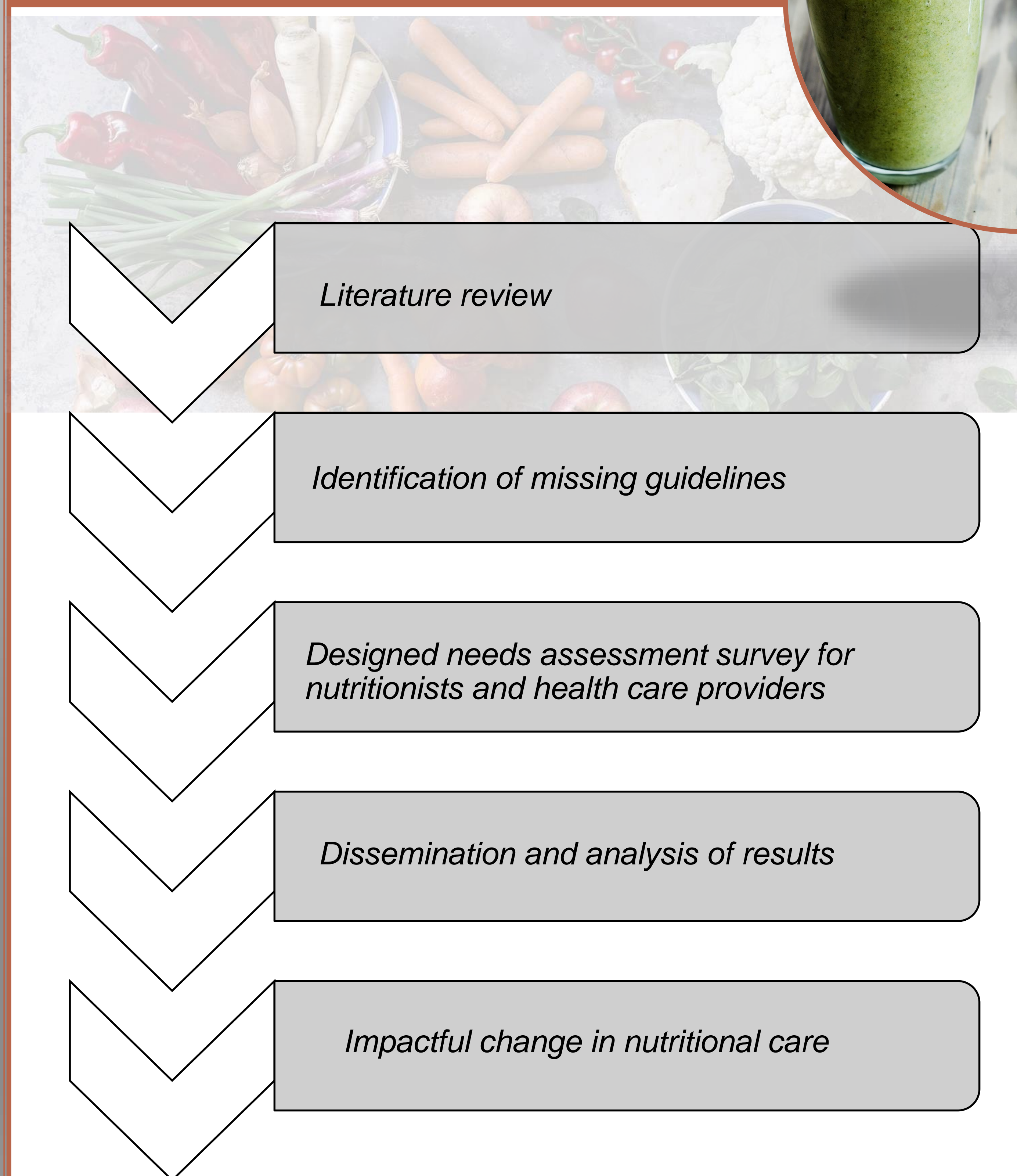
- Patients with metabolic diseases often have unique nutritional needs that can greatly affect their daily lives
- Limited information for caretakers and health care providers presents a challenge in the care of these patients
- Definitive nutrition guidelines for both health care providers and caretakers need to be developed



METHODS

- We conducted a PubMed literature review for nutrition care needs in pediatric patients with rare disease
- We analyzed the literature to better understand current clinical practices in nutrition care
- We identified gaps in nutrition care practices and areas for further improvement and development
- Using our findings, we created a needs assessment survey consisting of 22 questions
- We will be distributing this survey to clinical nutritionists and other health care providers who serve this population

NEEDS ASSESSMENT



SURVEY

Our survey consists of 22 questions in both free-response and multiple-choice formats. The questionnaire aims to gain an understanding of current practices, identify opportunities for improvement, and gather recommendations for standardized guidelines in rare disease nutrition care.

Examples of questions included in the survey:

- What is the most impactful challenge you face with your patient population?
- What nutrient delivery methods do your patients rely on the most?
- Do you and/or the caretakers change nutritional care based on palliative care reasons?
- If you could change one aspect of nutritional care in this patient community, what would that be?

EXPECTED OUTCOMES

- The survey will highlight areas of ongoing need in nutrition care for pediatric patients with rare disease
- Survey responses may also reveal strategies and practice guidelines that have worked for this population, which can be shared more broadly with others in this field
- We plan to share our findings with key stakeholders and policymakers in the rare disease community
- Additional research and/or surveys may arise from the findings

LITERATURE / RESOURCES

Literature Review:
Figueroa MJ, Rojas et al. Morbimortality associated to nutritional status and feeding path in children with cerebral palsy. Rev Chil Pediatr. 2017;88(4):478-486.
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Nelms CL, et al. Assessment of nutritional status in children with kidney diseases-clinical practice recommendations from the Pediatric Renal Nutrition Taskforce. Pediatr Nephrol. 2021 Apr;36(4):995-1010.

Image sources: The GFPD, Novartis, and MS Office stock images

ACKNOWLEDGEMENTS

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