Evaluating Association of Nijmegen Paediatric CDG Rating Scale (NPCRS) with Patient Reported Outcome Measurement Information System (PROMIS) in Patients with Phosphomannomutase 2 Deficiency (PMM2-CDG)

Anab Mohamed[1], Anna N. Ligezka, MS[1], Eric C. Polley PhD[3], and Eva Morava MD, PhD[1,2]

[1]Department of Clinical Genomics, Mayo Clinic, Rochester, MN, USA
[2]Biochemical Genetics Laboratory, Department of Laboratory Medicine and Pathology, Mayo Clinic, Rochester, MN, USA
[3]Bioinformatics Core, Mayo Clinic, Rochester, MN, USA

Introduction & Objectives

➤ Patient-reported outcomes (PROs) measure important aspects of disease burden, however they have received limited attention in CDG research and clinical care, in particular PMM2-CDG [1].

➤ PROs consistently provide critical information regarding the patient experience and can be used to establish effective therapeutic interventions [1].

➤ Here, we aim to compare patient-reported outcomes experienced by PMM2-CDG patients using the Patient-Reported Outcomes Measurement Information System (PROMIS) tool with clinician assessment of disease severity using The Nijmegen Paediatric CDG Rating Scale (NPCRS).

Results

Figure 1
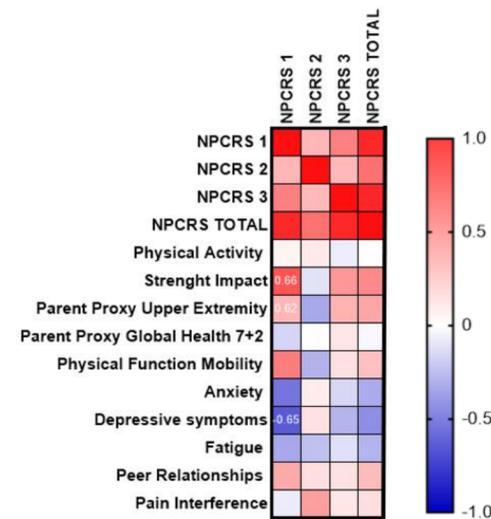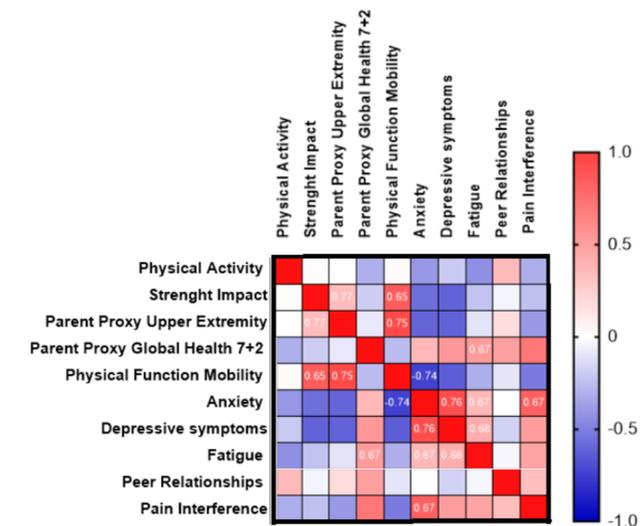
PROMIS vs. NPCRS

Figure 2

PROMIS vs. PROMIS

Methods

➤ A cohort of 26 PMM2-CDG parents of children completed the PROMIS questionnaire. Participants were clinically assessed by physicians using NPCRS to determine disease severity.

➤ The PROMIS questionnaire consisted of 10 general domains that assess physical activity, strength, mental, and social aspects of health. NPCRS consisted of 3 general domains: Section I (Current Function), Section II (System Specific Involvement), and Section III (Current Clinical Assessment).

➤ Pearson correlation analyses were used to determine the correlations between PROMIS and NPCRS and the correlations within the 10 PROMIS domains via use of a matrix correlation.

Results

➤ Three of the 10 PROMIS items (Strength Impact, Depression Symptoms and Parent Proxy Upper Extremity) were associated with NPCRS 1 section (Current Function) (**Figure 1**).

➤ Positive correlations ranged from r=0.62 for Parent Proxy Upper Extremity (P=0.01) to r=0.66 for Strength Impact Upper Extremity (P=0.04) (**Figure 1**).

➤ One negative correlation was found for Depressive Symptoms (r=-0.65, p=0.03) (**Figure 1**).

➤ There was no significant relationship between the NPCRS Section 2, 3, Total NPCRS scores and other PROMIS subscales.

➤ Within the 10 PROMIS domains showed that a total of nine subsections in PROMIS are correlated (**Figure 2**).

➤ Anxiety was positively correlated with Depressive Symptoms (r=0.76, p=0.002), Fatigue (r=0.67,p=0.022) and Pain Interference (r=0.67, p=0.051) and negatively correlated with Physical Function Mobility (r=-0.74, p=0.002) (**Figure 2**).

➤ Additionally, there was a relationship between Parent Proxy Upper Extremity and Physical Function Mobility (r=0.75, p=0.001) (**Figure 2**).

Conclusions

➤ There is an important need to assess how well clinical assessments of disease severity match with PROs in PMM2-CDG patients.

➤ NPCRS 1 has been shown to strongly correlate with patient assessment of 3 of the PROMIS subscales.

➤ PROs such as PROMIS provide important insight into the quality of life experiences by patients, thus PROs can improve the quality of patient care for PMM2-CDG patients by creating a holistic approach to clinical decision-making.

➤ Our findings suggest that there is a strong correlation between clinical and PRO reports. These findings are critical as they point to high agreement between physician and patient reports of disease burden and impacts on quality of life.

Reference

1. Overbeek CL, Nota SP, Jayakumar P, Hageman MG, Ring D. The PROMIS physical function correlates with the QuickDASH in patients with upper extremity illness. Clin Orthop Relat Res. 2015 Jan;473(1):311-7. doi: 10.1007/s11999-014-3840-2. Epub 2014 Aug 7. PMID: 25099262; PMCID: PMC4390943.