Background

Pyruvate kinase (PK) deficiency is an ultra-rare hemolytic anemia caused by autosomal recessive mutations in the PKLR gene.

An understanding of how signs and symptoms of pyruvate kinase (PK) deficiency can impact health-related quality of life (HRQoL) is important for optimal disease management and treatment of patients with PK deficiency.

Methods

- Primum: to develop PRO measures for symptom and impact assessment in PK deficiency according to the United States (US) Food and Drug Administration (FDA) guidance.
- Secundum: to compare the newly developed PROs to the EORTC QLQ-C30 and SF-36v2®.

Objective

- To develop PRO measures for symptom and impact assessment in PK deficiency, and to compare the newly developed PROs to the EORTC QLQ-C30 and SF-36v2®.

Results

- The initial draft of the Pyruvate Kinase Deficiency Impact Assessment (PKDI) was an 8-item PRO measure of common impacts of PK deficiency experienced by adults, using an 11-point scale and a 0–100 score.
- Following the cognitive interview, an item was added and another was significantly modified.
- The second version of the PKDI, following cognitive debriefing, consisted of 14 items measuring 12 concepts, with 2 skip pattern items.
- The revised conceptual framework for the PKDI is presented in Figure 2.

Discussion

- The newly developed PKDI and PKDA are more relevant and specific to the PK deficiency patient population.

Conclusions

- The newly developed PKDI and PKDA are more relevant and specific to the PK deficiency patient population.

Table 1: Comparison of conceptual coverage of PKDI and PKDA to EORTC QLQ-C30 and SF-36v2®

<table>
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<th>Measure</th>
<th>Emotional</th>
<th>Social</th>
<th>Functional</th>
<th>Physical</th>
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<td>Yes</td>
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<td>SF-36v2®</td>
<td>No</td>
<td>No</td>
<td>Yes</td>
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Figures

Figure 1. Overview of methods for PRO development

Figure 2. Revised conceptual framework for the PKDI

Figure 3. Revised conceptual framework for the PKDD

Figure 4. Comparison of other measurement characteristics of PKDD and PKDIA to EORTC QLQ-C30 and SF-36v2®