

Analyzing Accessibility and Suitability of Online Krabbe Disease Resources

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Background and Objective Between 25 and 30 million Americans are affected by rare diseases. Krabbe disease (KD) is a neurodegenerative leukodystrophy affecting approximately 1 in 100,000 births in the United States. KD has been screened for in select states since 2006 but was recently suggested for addition to the Recommended Uniform Screening Panel. Following a diagnosis, parents frequent the internet to learn about specific medical conditions and necessary medical follow-up. Because disparities in management and prognosis are often associated with health literacy levels, patient education materials (PEMs) must be accessible to their intended audiences. This study aimed to assess the accessibility and suitability of online KD resources, using results to provide recommendations for resource improvement.

Methods A Google search was conducted utilizing common search terms parents may use to identify patient-centered online KD resources. Twelve online KD resources were analyzed. These resources were compared against an author-developed list of essential information for patients and families. Readability was assessed using the Flesch-Kincaid Reading Grade Level and the Simple Measure of Gobbledygook Reading Grade Level, available through Readable.com. Suitability was measured by two reviewers utilizing the Suitability Assessment of Materials (SAM) Tool and the Patient Education Materials Assessment Tool (PEMAT) to reach a consensus assessment for each resource. Resources were classified as ‘superior’, ‘adequate’, or ‘not suitable’ based on the SAM Tool and rated on understandability and actionability using the PEMAT.

Results Most selected resources had readability grade levels above the recommended sixth to eighth grade reading levels for PEMs, ranging from 7.4 to 14.6. Eleven out of twelve (91.7%) resources were rated as ‘adequate’ or higher using the SAM Tool. PEMAT understandability scores ranged from 55.05% to 94.12%, and actionability scores ranged from 0% to 83.33%.

Conclusions While resources were easy to navigate, they struggled using common language to make the information understandable to the broader population, utilizing graphics appropriately, and promoting interactivity and presenting concrete next steps based on given information. Future resource development should focus on implementing action steps parents can take after diagnosis and improving readability by using common terminology and graphics to increase understanding.

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Preface

The basis of this research stemmed from my interest in neurodevelopmental disorders and passion for health literacy. I could not have developed my initial idea into this final project without a strong support system. I would like to take this opportunity to express my deep appreciation for everyone who supported me during this process.

Thank you to Jodie Vento for guiding me through the preliminary stages of my thesis and connecting me with individuals in the neurology and advocacy fields. Additional thanks to Curtis Weaver for his support with the Suitability Assessment of Materials Tool and Patient Education Materials Assessment Tool analysis.

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1.0 Introduction

Krabbe disease (KD) is a neurodegenerative leukodystrophy affecting approximately 1 in 100,000 births in the United States (Chapman et al., 2019; MayoClinicLaboratories, 2023). Also classified as a genetic disorder of metabolism, KD results from deficient galactocerebrosidase, a lysosomal enzyme, and an accumulation of psychosine and galactosylceramide in nerve cells (Bascou, DeRenzo, Poe, & Escolar, 2018). An absence of galactocerebrosidase activity, leading to neuronal death and subsequent deficient myelination in KD, causes slow or absent infantile and childhood development, spasticity of extremities, and seizures, among various other clinical features (J. J. Orsini et al., 2016). Late-onset KD, with some residual enzyme activity, results in spasticity with truncal hypotonia (decreased muscle tone affecting the trunk of the body), vision loss, seizures, and peripheral neuropathy. As the condition continues to progress, symptoms worsen, leading to premature death when untreated (Bascou et al., 2018).

Krabbe disease is currently included on the newborn screening (NBS) panels of some states in the United States. Newborn screening is a public health program aimed at identifying infants with an increased risk for genetic, endocrine, and metabolic conditions (ExpectingHealth, 2022). Rather than providing a diagnosis for these conditions, NBS alerts healthcare providers to infants who may benefit from early intervention and treatment after confirmatory testing. According to a study performed by Blackwell et al., a majority of families with a child with KD or with a family history of KD supported nationwide implementation of NBS for KD (Blackwell et al., 2020). Due to a strong push from families, KD was reviewed and recommended for addition to the Recommended Uniform Screening Panel in January 2024 (Eastman, 2024). However, parents also

expressed their concern for insufficient resources following diagnosis, which is important to address as screening and testing for KD expand (Blackwell et al., 2020).

Studies have shown parents consult online resources regarding their child's health for a number of reasons, with the primary motives being to learn about their child's diagnosis, available treatments, and specific associated medical conditions (Tozzi et al., 2013; Tuffrey & Finlay, 2002; Yardi, Caldwell, Barnes, & Scott, 2018). Online health information searches also aided in determining if children required additional care or answering remaining questions after an appointment (Yardi et al., 2018). While a majority of individuals reference the internet for health information, only 12 percent of adults living in the United States have proficient health literacy skills, defined as being able to read, comprehend, and use health information to take an active role in their health care ("Health Literacy in Healthy People 2030," 2020). Lower health literacy leads to worse health outcomes and more confusion with genetics-related information (Chapman et al., 2019; Shahid et al., 2022).

Taking this into account, the Agency for Healthcare Research and Quality (AHRQ) and The National Institutes of Health (NIH) recommend patient education materials (PEMs) be written between sixth and eighth grade reading levels (Agency for Healthcare Research and Quality, 2020; Rooney et al., 2021). Previously validated tools can be used to analyze readability and suitability of PEMs. The quality and appropriateness of online resources for various medical conditions, like glaucoma, coronavirus disease 2019, and carcinoma, for example, have been previously investigated, and many are reported to be written well above the recommended sixth and eighth grade reading levels (Crabtree & Lee, 2022; Georgsson & Carlsson, 2022; Tan, Ko, & Fan, 2023). At this time, no studies have assessed the resources parents may encounter when seeking information on KD or related search queries (e.g., globoid cell leukodystrophy).

This project reviewed internet search results related to KD and analyzed resources that are targeted toward parents with a child diagnosed with KD. The search was performed through Google Chrome using a set of search terms that the authors determined are most likely for parents to use when gathering information.

1.1 Specific Aims

1. Assess the readability of online resources for Krabbe disease using the Flesch-Kincaid and SMOG Reading Grade Levels and compare these to the recommended sixth to eighth grade reading levels for health-related materials as outlined by the Agency for Healthcare Research and Quality and the National Institutes of Health
2. Evaluate the accessibility and suitability of online Krabbe disease resources for families by utilizing the SAM Tool and PEMAT, and comparing resource content against a specified list of content by common sections
3. Develop recommendations for parent Krabbe disease education resource development/improvement based on findings from Specific Aims 1 and 2

2.0 Manuscript

2.1 Background

Rare diseases collectively affect between 25 million and 30 million Americans. Krabbe disease (KD), also called globoid cell leukodystrophy, is a rare neurodegenerative leukodystrophy caused by an inborn error of metabolism (Bascou et al., 2018; Chapman et al., 2019; MayoClinicLaboratories, 2023). Galactocerebrosidase is a lysosomal enzyme responsible for clearing the body of psychosine and galactosylceramide (Bascou et al., 2018). Deficient galactocerebrosidase results in accumulation of these substrates in nerve cells. Elevated psychosine levels are toxic to oligodendrocytes and Schwann cells, two cell subtypes responsible for myelination in the central and peripheral nervous systems, respectively (Suzuki, 2003). Galactocerebrosidase deficiency leads to neuronal death and insufficient myelination, causing slow or absent infantile and childhood development, spasticity of extremities, and seizures, among other characteristic features of KD (J. J. Orsini et al., 2016).

KD is characterized into two main types, primarily based on time of symptom onset: infantile-onset KD and late-onset KD (Table 1). Infantile onset refers to patients with onset of symptoms in the first year of life and is the most aggressive and devastating form of KD (Duffner et al., 2009). Individuals with this form of KD typically develop normally in the first few months of life with subsequent severe, rapid deterioration. Without treatment, children with KD can develop complications as early as the neonatal period (Bascou et al., 2018). The current standard of care for KD is hematopoietic stem cell transplantation (HSCT); however, clinical trials are underway for other interventions including gene therapy and administration of intrathecal

umbilical cord blood derived oligodendrocyte-like cells (Forge Biologics, 2023; Kurtzberg, 2023). The average lifespan of untreated individuals with infantile-onset KD is two years; however, some individuals with infantile-onset KD have lived until nine years of age (J.J. Orsini, Escolar, Wasserstein, & Caggana, 2018). Late-onset KD describes individuals who initially present with symptoms after twelve months of age, with some individuals not presenting until the seventh decade of life (Hagberg, Sourander, & Svennerholm, 1963). This phenotype tends to be less severe and more slowly progressing (Suzuki, 2003). Late-onset KD is further divided into three subtypes: late-infantile onset, juvenile onset, and adult onset. Generally, the earlier the age of onset, the more severe the phenotype and prognosis.

Table 1 Common Symptoms of Infantile-Onset and Late-Onset Krabbe Disease

Infantile-onset KD (≤ 12 months)	Late-onset KD (> 12 months)
<ul style="list-style-type: none"> • Excessive crying • Irritability • Gastrointestinal issues • Spasticity and fisting • Absent tendon reflexes • Staring episodes • Peripheral neuropathy 	<ul style="list-style-type: none"> • Slow motor development • Spasticity with truncal hypotonia • Vision loss • Esotropia • Seizures • Peripheral neuropathy
(Krabbe, 1916; J.J. Orsini et al., 2018)	

KD diagnosis is initiated via one of two avenues: clinical findings in a symptomatic proband or low galactocerebrosidase activity followed by second-tier test showing an elevated psychosine level of ≥ 10 nM in an asymptomatic infant via newborn screening (NBS) that leads to confirmatory diagnostic testing. While KD was initially not included on the Recommended Uniform Screening Panel (RUSP), which is maintained by the Secretary of the Department of Health and Human Services, when proposed in 2023 due to knowledge gaps about and risks of

HSCT (Federal Advisory Committee, 2023), some states implemented NBS for this condition anyway.

Blackwell and colleagues previously conducted a survey to explore family perspectives regarding inclusion of KD on NBS. Ninety-seven percent of participants supported inclusion of KD on NBS, citing reasons such as presymptomatic detection, available treatment, and the ability to build on scientific knowledge to benefit families in the future. Those who did not support adding KD on NBS cited a lack of follow-up protocol, no curative treatment, and vague information received at the time of diagnosis (Blackwell et al., 2020).

In January 2024, KD was once again proposed for addition to the RUSP, and due to overwhelming support from affected families, the U.S. Advisory Committee on Heritable Disorders in Newborns and Children voted 10-3 to recommend addition of KD to the RUSP (Eastman, 2024). Because the RUSP is a national guideline for inherited disorders which should be included on states' NBS panels, states that have not yet implemented screening for KD will likely follow this recommendation. Thus, it is imperative online information about KD be accurate, suitable, and accessible for parents whose child is diagnosed with this genetic condition.

According to the *Global Statshot Report*, nearly 60 percent of people worldwide use the internet to find information (Kemp, 2023). The subset of individuals using the internet for health-related information is rising (Bianco, Zucco, Giuseppe A Nobile, Pileggi, & Pavia, 2013; Kubb, Foran, Scott, & Volkman, 2020). As more individuals receive a diagnosis of KD, either by NBS or clinical criteria, it is likely this trend for online health information will continue in families with KD. When it comes to their children, parents consult online resources primarily to learn about specific diagnoses and treatments, to determine if their child needs additional care, and to prepare for and follow up from doctors' visits (Nicholl, Tracey, Begley, King, & Lynch, 2017; Tozzi et

al., 2013; Tuffrey & Finlay, 2002; Yardi et al., 2018). However, online information is often not accessible to the general public. For example, online patient education materials (PEMs) for conditions such as glaucoma and carcinoma that were analyzed for quality and readability reported that these materials were written well above the Agency for Healthcare Research and Quality (AHRQ) and National Institutes of Health (NIH) recommended sixth to eighth grade reading levels (Crabtree & Lee, 2022; Georgsson & Carlsson, 2022; Tan et al., 2023).

Studies have shown the average individual in the United States reads at an eighth-grade level or lower (Safeer & Keenan, 2005). Only 12 percent of adults living in the United States have proficient personal health literacy skills ("Health Literacy in Healthy People 2030," 2020; Rikard, Thompson, McKinney, & Beauchamp, 2016). According to Healthy People 2030, personal health literacy is "the degree to which individuals have the ability to find, understand, and use information and services to inform health-related decisions and actions for themselves and others" ("Health Literacy in Healthy People 2030," 2020). Health literacy encompasses not only the ability to understand health information but also the capacity to use that information to make knowledgeable decisions about health and treatment (Ratzan, 2001). Low health literacy is a universal issue, not confined to one class of individuals. Szabo, Biro, and Kosa analyzed comprehension of PEMs among laypersons, non-professional healthcare workers (individuals working in healthcare who do not require professional qualification, like technicians or medical administrative assistants), and medical students (Szabó, Bíró, & Kósa, 2021). The researchers discovered the laypersons' accuracy rate was no better than randomly selecting answers, and the non-professional healthcare workers were not much better. Even medical students scored only slightly better than chance on questions related to insurance claims and ethical issues (Szabó et al., 2021).

The impacts of lower health literacy lead to worse health outcomes (Shahid et al., 2022). Patients with low health literacy are more likely to have higher rates of hospitalization, lower treatment compliance, and less overall involvement in their own health care (Safeer & Keenan, 2005; Shahid et al., 2022). Likewise, there are implications of low health literacy in regard to genetic testing. Miyoshi and Watanabe found that individuals with lower health literacy had more difficulty understanding the risks and benefits when a new genetic test was presented to them than individuals with high health literacy (Miyoshi & Watanabe, 2023). Furthermore, Kaphingst et al. investigated the relationship of factors like health literacy, education, and empirical melanoma risk with responses to genetic testing. Their study showed a negative correlation between health literacy and confusion during and after genetic results disclosures (Kaphingst et al., 2021). Beyond simply understanding what a test entails, studies show individuals with low health literacy are less willing to apply genetic testing results to personal health decisions (Chapman et al., 2019). A greater proportion of individuals with high health literacy are willing to undergo genetic testing and use that information to inform their health decisions (Chapman et al., 2019).

To reduce health disparities, PEMs should be tailored to the intended audience. Assessing readability and suitability can help determine the overall appropriateness of a resource. Readability is defined as “the quality of being easy and enjoyable to read” (Cambridge Dictionary, 2023). The AHRQ and NIH recommend PEMs be written between a sixth and eighth grade reading level (Agency for Healthcare Research and Quality, 2020; Rooney et al., 2021). Suitability is defined as “the fact of being acceptable or right for something or someone” (Cambridge Dictionary, 2023).

Healthcare professionals and researchers can use previously validated tools to analyze readability and suitability of PEMs. Readability assessments have been performed on PEMs for

various health conditions, most often using the Flesch-Kincaid Index and Reading Ease Scale, and the Simple Measure of Gobbledygook (SMOG) scale ("Improving Health Literacy One Word at a Time," 2006; Kher, Johnson, & Griffith, 2017; Morony, Flynn, McCaffery, Jansen, & Webster, 2015; Prabhu et al., 2016). Several studies have assessed the accessibility and suitability of online education materials for their respective audiences (Fortuna, Riddering, Shuster, & Lopez-Jeng, 2020; Martin et al., 2022; Smith et al., 2014; Tian, Champlin, Mackert, Lazard, & Agrawal, 2014). The majority of online PEMs were well below the recommended suitability score of 70 percent. Using the SAM Tool, Fortuna et al. determined the average suitability score for PEMs targeted to people with age-related macular degeneration was 53 percent, which translates to a rating of 'adequate' (Fortuna et al., 2020). In a study regarding PEMs for patients undergoing elective surgery for colorectal cancer, 9.6 percent were ranked 'superior', 76.8 percent were rated 'adequate', and 13.6 percent were considered 'not suitable' (Smith et al., 2014). Tian et al. found a majority of PEMs for colorectal cancer patients to be 'adequate' with regard to graphics, layout, and learning stimulation; however, these resources did not meet the informational demands of patients considering surgery (Tian et al., 2014). The aforementioned studies encapsulate only a small proportion of research on suitability and accessibility of online PEMs; however, the overall trend is these materials often fall short of meeting the suggested criteria.

This study aimed to explore online KD resources through clinical and public health lenses, assessing readability, suitability, and accessibility. Based on these results for the online KD resources, the researchers developed recommendations for parent education resource improvement to better educate parents and encourage proper management of KD. Improved accessibility and suitability of online resources for parents have the potential to result in better comprehension and preparedness of parents when speaking with providers, leading to enhanced overall care for

children diagnosed with KD. Furthermore, with KD being highly discussed in NBS legislation, understandable online KD resources could benefit policymakers in building a foundational knowledge as well.

2.2 Methods

The University of Pittsburgh Institutional Review Board determined that this research study is not human subjects research as shown in Appendix A.

2.2.1 Collection of Resources

An internet search performed in January 2024 using a Google Chrome Incognito tab yielded the webpages listed in Appendix B. Using an Incognito tab inhibits the browser from remembering previous search history, reducing potential search engine bias. Five search terms were used to generate the list of webpages: “Krabbe disease”, “globoid cell leukodystrophy”, “parent resources for Krabbe disease”, “GALC deficiency”, and “Krabbe disease newborn screening.” The top ten webpages were kept for each search term (Appendix B).

Resources for analysis were chosen based on their target audience. The audience of interest for this study was parents whose child(ren) screen(s) positive for or is/are confirmed to have Krabbe disease. A resource’s target audience was determined based on a website’s “About Us” section. Where not explicitly stated, the authors of this paper determined a resource’s implied intended audience. Twelve resources were chosen for analysis (Table 2).

Table 2 Search Term(s) that Returned Analyzed Resources

Resource	Search Term(s)
Organization A	Krabbe disease, Globoid cell leukodystrophy, Parent resources for Krabbe disease
Organization B	<i>Chosen by authors/family advocate recommendation</i>
Organization C	<i>Chosen by authors/family advocate recommendation</i>
Organization D	Parent resources for Krabbe disease, GALC deficiency, Krabbe disease newborn screening
Organization E	Parent resources for Krabbe disease
Organization F	Parent resources for Krabbe disease
Organization G	Krabbe disease newborn screening
Organization H	<i>Chosen by authors/family advocate recommendation</i>
Organization I	Krabbe disease, Globoid cell leukodystrophy
Organization J	<i>Chosen by authors/family advocate recommendation</i>
Organization K	Krabbe disease newborn screening
Organization L	Krabbe disease, Globoid cell leukodystrophy, GALC deficiency

Analyzed resources included those that provided information on KD for parents with child(ren) with KD (Figure 1). A few search results discussed globoid cell leukodystrophy in animals; these were not included in the analysis. Resources targeted toward providers (e.g., GeneReviews) were also excluded from analysis. Additionally, online resources produced by individual hospitals or hospital systems were excluded, as this would primarily represent hospitals near Pennsylvania, Ohio, and Maryland, skewing representation.

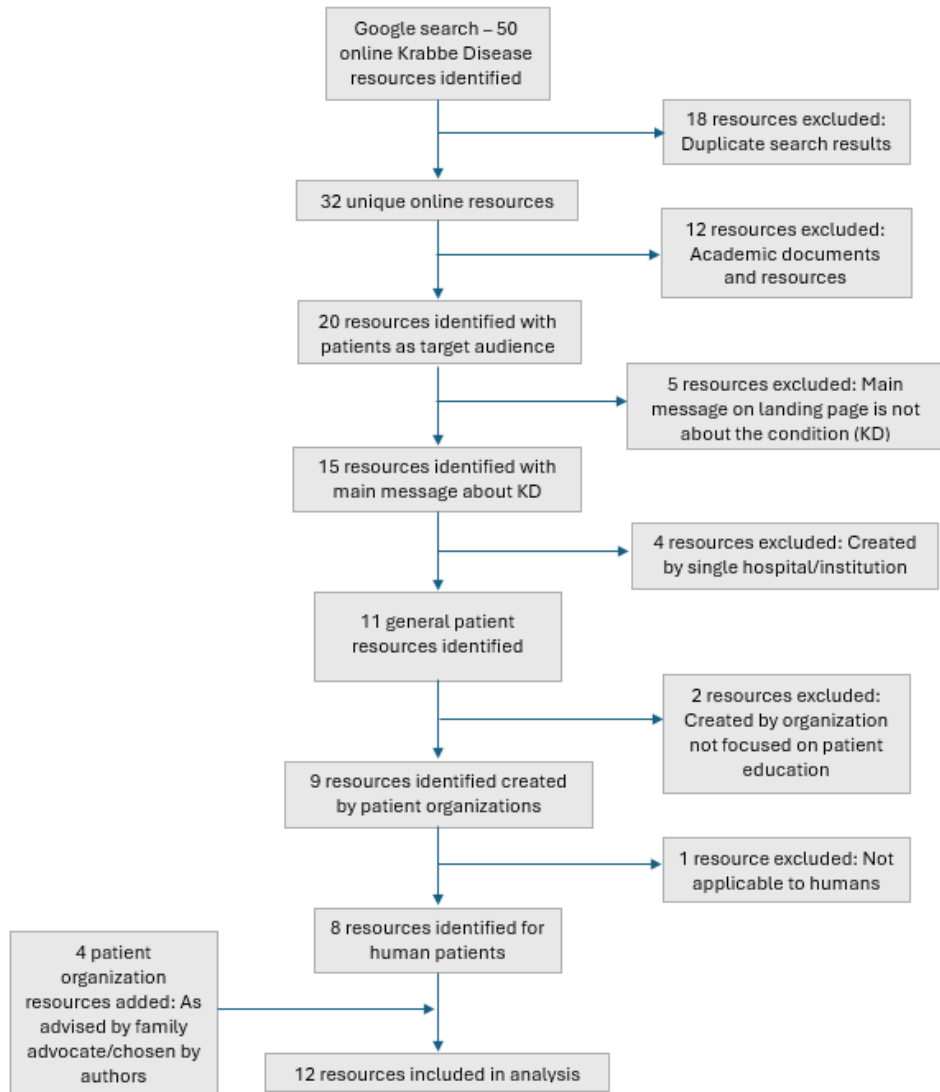


Figure 1 Flow Chart for Resource Identification, Inclusion and Exclusion Criteria. This flow chart illustrates the database used for this study, the number of search results, and the measures used for exclusion.

2.2.2 Accessibility Assessment by Content Criteria

A list of criteria regarding important content of materials for families impacted by KD was developed by the authors of this paper. To obtain as high a level of objectivity as possible, each website was determined to either include or not include each criterion. The criteria included: a

basic description of KD, KD symptoms, genetics of the condition, genetic counseling, treatment options, clinical trials, support groups/resources, and next steps.

2.2.3 Readability Scores

Readability of written health information can be assessed using various formulas. The Flesch-Kincaid Reading Grade Level was created to analyze technical material but is now recognized for its reliability at determining readability of a wide range of written documents. The Flesch-Kincaid Reading Grade Level determines readability based on average sentence length and the number of syllables per word (Table 3) (Jindal & MacDermid, 2017). The tool measures readability between a grade 5 level and college level and provides a score that corresponds to a U.S. grade level (Jindal & MacDermid, 2017). A limitation of the Flesch-Kincaid Reading Grade Level is its underestimation of grade level due to recognition of every period (including decimals, bullets, and abbreviations) as the end of a sentence.

The Simple Measure of Gobbledygook (SMOG) Reading Grade Level, also known as the SMOG Index, is another readability tool proven reliable for a range of texts, including healthcare materials. The tool calculates the number of education years needed to understand the text by taking into account the number of sentences and the number words with three or more syllables (Table 3).

Table 3 Readability Tools and Corresponding Formulas

Assessment tool	Formula
Flesch-Kincaid Reading Grade Level	$(0.39 \times \text{average number of words per sentence}) + (11.8 \times \text{average number of syllables per word}) - 15.59$
SMOG Reading Grade Level	$1.0430 * \sqrt{(\# \text{ of polysyllabic words}) * (30 / \# \text{ of sentences})} + 3.1291$

Readability scores were calculated using Readable.com. Among other functions, Readable.com is an online tool that determines the readability of a text using various formulas: Flesch-Kincaid, Gunning-Fog, SMOG, Coleman Liau, and more. Text from the main landing page of each resource was copied into the Readable.com text box. Links and sidebar information were not included in the analysis. The Flesch-Kincaid Reading Grade Level and SMOG Index were reported for each resource in this study.

2.2.4 Suitability Assessment

Suitability assessment followed a similar format as previous studies, utilizing validated tools including the Suitability Assessment of Materials (SAM) Tool and the Patient Education Materials Assessment Tool (PEMAT) (Crabtree & Lee, 2022; Fortuna et al., 2020; Georgsson & Carlsson, 2022; Kher et al., 2017).

The SAM Tool was created by Doak et al. during an adult health education project under the Johns Hopkins School of Medicine and was designed to assess suitability of educational health resources, as well as appropriateness across cultures (Doak, Doak, & Root, 1985). The tool considers factors affecting readability and comprehension including content, literacy demand, graphics, layout and typography, learning stimulation and motivation, and cultural appropriateness (Doak et al., 1985). Each factor is awarded a score of zero, one, or two, based on standards associated with each score, which can be found on the scoresheet. Individual factor scores are totaled, which is then divided by the maximum score possible, leaving researchers with a percent interpretation (Table 4). SMOG Reading Grade Levels were incorporated into the scoring for the SAM Tool, as the SMOG formula has been shown to be the best measure of health information readability (Wang, Miller, Schmitt, & Wen, 2013).

The PEMAT was developed by Shoemaker, Wolf, and Brach in collaboration with the AHRQ (Agency for Healthcare Research and Quality, 2020). Its purpose is to evaluate the understandability and actionability of patient education materials, presenting scores as a percentage, so they can be compared between resources. This tool incorporates content, word choice and style, use of numbers, organization, layout and design, and use of visual aids into the understandability analysis. For actionability, the tool factors in how many actionable steps are explicitly stated in the material, if actions are broken into manageable steps, and if/how a resource explains how to use visual aids or perform calculations. Two versions of the PEMAT exist: printable (for brochures, pamphlets, and printable websites) and audio/visual (for multimedia materials). This allows for more accurate analysis of patient education materials that include videos and pictures, where instruments like the SAM Tool are limited.

In this study, two reviewers analyzed each resource individually using the SAM Tool and PEMAT scoresheets for consistency. Screenshots of all resources were used in case the websites changed during the analysis period or if reanalysis was needed. All scores were determined during January 2024. The reviewers met after completing analysis to discuss scores and came to an agreement on final interpretation for most resources. For any SAM Tool or PEMAT scores resulting in discordant interpretations (superior/adequate, adequate/not suitable), a third reviewer analyzed the respective resource. The third reviewer's score was the deciding factor for a resource's interpretation and was incorporated into the average suitability score for each assessment tool. For example, if the first two reviewers rated the resource as 'adequate' and 'superior', and the third reviewer scored 'adequate', the resource was considered to be 'adequate'. All factors were double-coded, apart from readability, which the primary author of this study assessed using Readable.com. All resources were analyzed using the SAM Tool, and eleven out

of twelve resources were analyzed using the PEMAT-Printable version. One resource was part text, part audio/visual. Thus, PEMAT-Printable version and PEMAT-Audio/Visual version were both used for analysis.

Table 4 Suitability Assessment Tools and Interpretation

Assessment tool	Interpretation
SAM Tool	Ranges from 0-100 0-39: not suitable material 40-69: adequate material 70-100: superior material
PEMAT	Ranges from 0-100 Higher percentage indicates greater understandability or actionability of a material

2.3 Results

The top ten results from each search term were noted for a total of 50 websites. The Google searches yielded 32 unique search results. From the search results, approximately 62 percent (20 out of 32) of websites were applicable for the target audience of parents with child(ren) with KD. The proportion of appropriate resources for parents was dependent on the search term. For example, “GALC deficiency” resulted in more challenging, peer-reviewed articles and resources for providers than “Krabbe disease”. Twelve online KD resources were chosen for accessibility and suitability analysis (Figure 1).

2.3.1 Content Criteria

The content of resources chosen for analysis was examined (Table 5). Only information on the landing page was included in the analysis. Many sites may have additional information about support groups and genetic counseling on other pages of the website; however, these pages were not analyzed in this study. All resources included a basic description of KD and mentioned the symptoms and genetics (e.g., inheritance pattern) of the condition. Only four out of twelve (33.3%) resources mentioned genetic counseling and/or how to find a genetic counselor with whom to speak about the child's condition. Rather than explicitly listing current clinical trials, most resources contained information related to this criterion in links to locate the clinical trials (e.g., ClinicalTrials.gov). Treatment for KD is developing, so information varied widely across resources. Eight resources discussed bone marrow transplant/hematopoietic stem cell transplant and medications for symptom management, while Organization A and Organization B mentioned broadly that there is no cure for KD. One resource, created by Organization E, included all content criteria.

Table 5 Content Inclusion by Resource

Resource	Basic Description	Symptoms	Genetics	Genetic Counseling	Treatment Options	Clinical Trials	Support Groups/ Resources	Next Steps	Percent of Topics Present (%)
Organization A	x	x	x		x	x	x		75
Organization B	x	x	x	x	x	x			75
Organization C	x	x	x		x				50
Organization D	x	x	x	x	x		x	x	87.5
Organization E	x	x	x	x	x	x	x	x	100
Organization F	x	x	x		x	x	x		75
Organization G	x	x	x		x				50
Organization H	x	x	x		x				50
Organization I	x	x	x			x	x		62.5
Organization J	x	x	x			x	x		62.5
Organization K	x	x	x		x		x		62.5
Organization L	x	x	x	x	x	x	x		87.5

2.3.2 Readability

Largely, the readability levels of the chosen online resources were well above the suggested sixth to eighth grade reading levels outlined by the AHRQ and the NIH (Figure 2). The average Flesch-Kincaid Reading Grade Level was 10.6, and the average SMOG Reading Grade Level was 12.5. The resource created by Organization F was closest to the recommended readability level, with a Flesch-Kincaid Reading Grade Level of 7.4 and SMOG Reading Grade Level of 9.8. Organization B and Organization L had the highest reading grade levels.

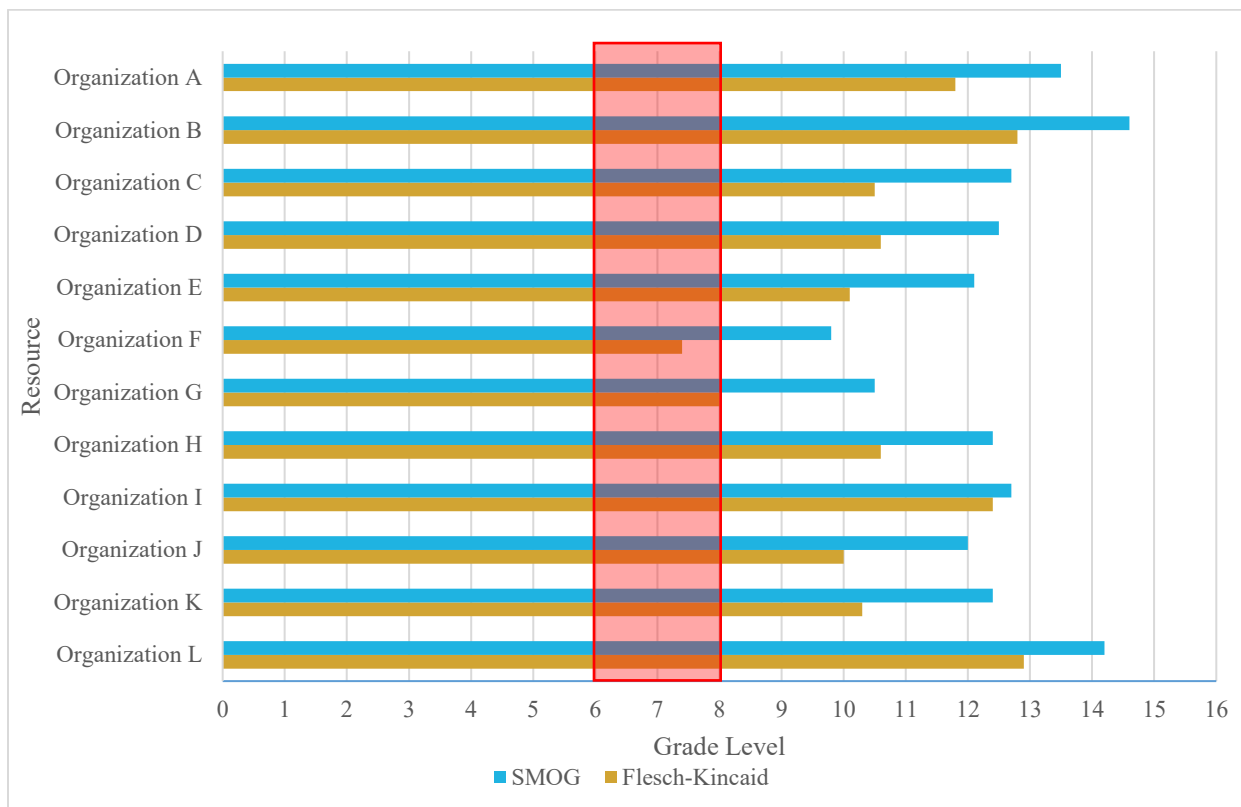


Figure 2 Resource Readability Scores. The (red) shaded region is the recommended sixth to eighth grade reading levels.

2.3.3 SAM Tool Results

Resources with scores of 0 to 39 percent were considered ‘not suitable’. From 40 to 69 percent, resources were ‘adequate’, and resources scoring 70 percent or higher were considered ‘superior’. Ten out of twelve resources (83.33%) scored ‘adequate’ on the SAM Tool (Appendix E). One resource, created by Organization E, was rated ‘superior’, with scores of 77.3 percent and 84.1 percent from Reviewers A and B, respectively. Except for one KD resource, the websites targeting parents of children with KD were appropriate for the population (Figure 3). The website created by Organization K scored 38.1 percent and 38.6 percent from Reviewers A and B, respectively, resulting in an interpretation of ‘not suitable’.

2.3.4 PEMAT Results

Understandability scores are calculated to determine how easily understood a resource is by the general population. The actionability score determines how actionable a resource is, based on tangible next steps from the information presented. The higher the percentage, the more understandable or actionable the resource is. For example, a resource with an understandability score of 90 percent is more easily understood by the general population than a resource with an understandability score of 50 percent. Scores of 70 percent or higher were considered ‘adequate’. Below that threshold, resources were considered ‘not suitable’.

Understandability scores ranged from 53.85 percent to 94.12 percent, and actionability scores ranged from 0 percent to 83.33 percent (Figure 3, Appendix F). Overall, the average understandability score was 69.83 percent, and the average actionability score was 26.76 percent.

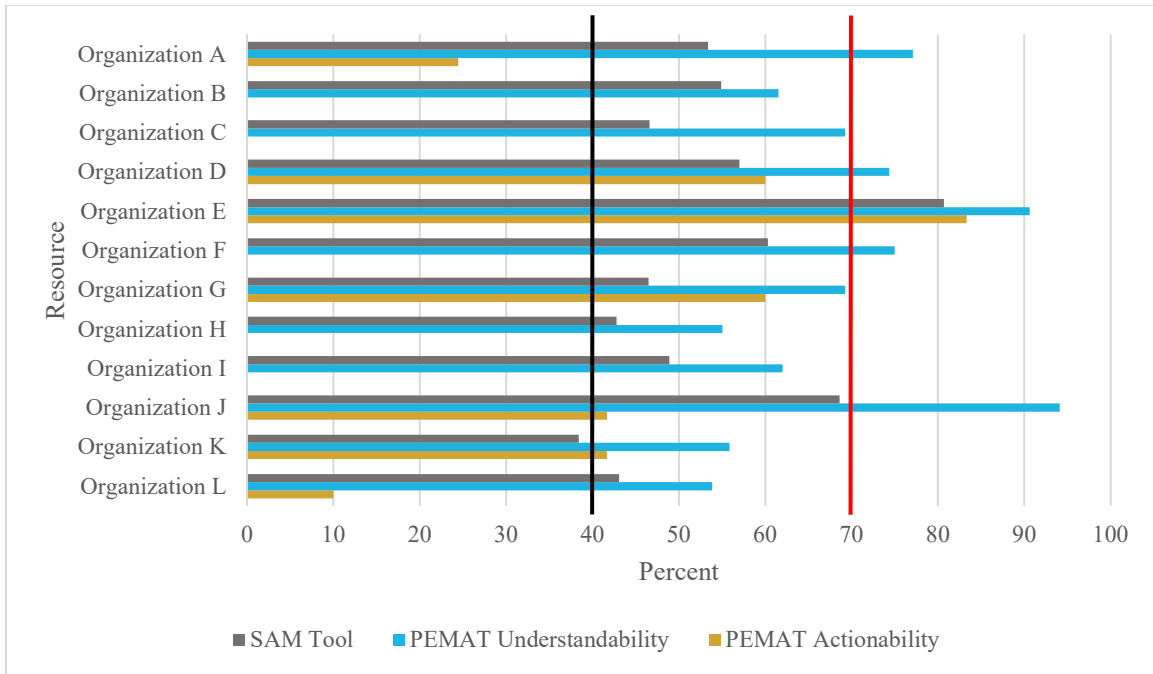


Figure 3 Resource Suitability Scores. The vertical (red) line at 70 represents the minimum superior level for the SAM Tool and the minimum adequate level for the PEMAT. The vertical (black) line at 40 represents the minimum adequate level for the SAM Tool. For resources with no third (gold) bar, PEMAT actionability score was 0 percent.

Table 6 Compiled Scores for All Resources

Resource	Flesch-Kincaid	SMOG	SAM Tool (%)	PEMAT Understandability (%)	PEMAT Actionability (%)
Organization A	11.8	13.5	53.4	77.08	24.44
Organization B	12.8	14.6	54.9	61.54	0
Organization C	10.5	12.7	46.6	69.23	0
Organization D	10.6	12.5	57.0	74.36	60.00
Organization E	10.1	12.1	80.7	90.63	83.33
Organization F	7.4	9.8	60.3	75.00	0
Organization G	8.0	10.5	46.5	69.23	60.00
Organization H	10.6	12.4	42.8	55.05	0
Organization I	12.4	12.7	48.9	62.02	0
Organization J	10.0	12.0	68.6	94.12	41.67
Organization K	10.3	12.4	38.4	55.88	41.67
Organization L	12.9	14.2	43.1	53.85	10.00

2.4 Discussion

Online resources are a convenient way for parents to quickly gain information about KD after a child's diagnosis. Nearly 75 percent of Americans who use the internet search for health-related information; yet, research consistently shows patient education materials are not easily understood by the general population, particularly those with lower health literacy (Smith et al., 2014; Tian et al., 2014; Wong, Saivasegaran, Choo, & Nah, 2018). As KD is added to more states' newborn screening panels, it is imperative online resources are understandable to parents, so they can make informed decisions about their child's care. In light of previous research conclusions regarding PEMs (Georgsson & Carlsson, 2022; Smith et al., 2014; Szabó et al., 2021; Tian et al., 2014) and the recent recommendation for KD to be added to the RUSP (Eastman, 2024), this study evaluated online KD resources based on content, readability, and suitability.

Criteria analysis showed most resources did not fully meet the informational needs of parents whose child is diagnosed with KD. While all resources included a basic description of KD, symptoms of the condition, and general genetics concepts (e.g., inheritance), many lacked discussions of treatment, next steps, and support groups. Studies show that, based on the Health Belief Model, the latter topics may enhance perceived benefits of treatment and health care and increase health motivation (Tian et al., 2014). Addressing concerns for accurate information and support and presenting potential solutions (e.g., treatment) is essential to increase parents' initiative using tangible resources (Tian et al., 2014).

Previous research has also shown there is a gap in genetics knowledge for practicing physicians. Although more recent graduates receive more genetics training in medical school, 63 percent of practicing physicians report feeling inadequately prepared or uncomfortable using genetic information (French, Kader, Young, & Fontes, 2023). Genetic counselors, however, have

expertise in explaining genetics jargon and navigating emotionally charged situations with families. Even so, many PEMs, surprisingly, did not include information regarding genetic counseling, which could help patients to find providers who are trained in this area.

Similar to previous studies that suggest patient health education materials are not accessible to the general population, this study showed KD resources have reading grade levels that are consistently above that of the AHRQ and NIH recommended sixth to eighth grade reading levels. Resources created by Organization F and Organization G met the recommended sixth to eighth grade reading levels based on the Flesch-Kincaid results but not the SMOG results. Flesch-Kincaid Reading Grade Levels for all KD resources were consistently lower than SMOG Reading Grade Levels. Previous studies show the SMOG Reading Grade Level is the best readability measure of health information when compared to other readability formulas (Wang et al., 2013). In these previous studies, the SMOG formula performed assessments with the highest level of consistency and was noted to have a higher level of expected comprehension, which is necessary for health-related materials. Understanding the main message of non-health-related materials is often sufficient, while it is essential that health-related materials are thoroughly understood to help ensure positive health outcomes (Wang et al., 2013).

Readability is a key component of creating a clear patient resource; however, it is not the only consideration. In fact, this study found resources with high reading grade levels (>11) may still be deemed 'adequate', or even 'superior', depending on other elements of the material. Results of analysis using the SAM Tool and PEMAT showed that most online KD resources for parents are adequately suited for their intended audience. Over 90 percent of resources analyzed in this study were rated 'adequate' or above using the SAM Tool, meaning most online resources for KD are appropriate for KD patients and parents. Approximately half of the resources analyzed

were considered ‘adequate’ based on the PEMAT understandability score. Although the resources generally met suitability standards, the findings of this study were consistent with those of previous studies, showing PEMs are not optimally created (Crabtree & Lee, 2022; Fortuna et al., 2020; Martin et al., 2022; Tan et al., 2023).

The suitability analysis highlighted several areas of weakness for PEMs: readability, interactivity where appropriate, use of graphics, and application-based information. Interactive content, such as “Question and Answer”, has been found to result in rigorous cognitive processing and enhanced memory of information (Xu & Sundar, 2016). Organizations could add interactive patient journey mapping to websites to create a more personalized experience for parents trying to determine the next best step. Few resources presented tangible steps for parents (e.g., contacting a genetic counselor, talking with a child’s pediatrician about further testing). In an emotionally charged situation, explicit instructions on “what to do when…” is imperative to patient empowerment and improvement of health outcomes. For example, websites could include a list of questions parents of KD patients may want to ask their child’s primary care provider. A list of specialists, including genetic counselors, with whom parents follow up could also help parents navigate which providers to reach out to if they do not have an established primary care physician for their child.

PEMAT analysis highlighted additional specific areas in need of improvement: summary, visual aids, explicit purpose, and common language. Similar to the SAM Tool, the PEMAT showed that online KD resources do not take advantage of visual aids. Visuals and common language can reduce the comprehension difficulty of the material (Guo, Zhang, Wright, & McTigue, 2020), so adding graphics to explain inheritance patterns, for example, may make the information more accessible. Resources also lacked summaries and explicit purpose statements.

The average person spends 10 to 20 seconds on a website before deciding if they will read more ("Average time spent on your website: web design," 2020); adding purpose statements and summaries of the resource's content provides readers the opportunity to glean the most important information in a succinct manner and decide if the website will be useful. Although not all SAM Tool and PEMAT interpretations were concordant, the overall trends of 'adequate' ratings remained.

PEMAT actionability scores were poorer than understandability scores, with only two resources being rated as 'adequate'. PEMs lacked refinement of readability and use of common language, interactive content, use of graphics and visual aids, and application-based information/modeled behaviors. Interactive components may not be feasible or appropriate for all content; however, creators of PEMs should focus on inclusion of appropriate graphics and visual aids to assist in comprehension and utilization of complex genetics and medical concepts. For example, a living chart of active clinical trials, in conjunction with questions to help families decide whether to participate in a trial, may be useful, as many websites simply had a link to clinicaltrials.gov. Such visual aids could also add much-needed behavior-modeling and mapping to help parents anticipate their next steps in the medical journey. Furthermore, based on the findings of this study, adjusting the language to meet the recommended sixth to eighth grade reading levels could improve overall interpretation. Organizations should focus on using phrases such as "damage of white matter in the brain that gets worse over time" rather than "progressive demyelination".

Table 7 Summary of Recommendations for Krabbe Disease Resource Improvement

The following are recommendations for Krabbe Disease resource improvement based on the findings of this study:
<ul style="list-style-type: none">• Explicit purpose statements and summary of resource content• Increased use of common language• Graphics where appropriate (e.g., inheritance pattern)• Patient journey mapping/interactive material• Suggestions for next steps parents can take

2.4.1 Limitations

The main limitation of this study is the lack of established protocol for analyzing PEMs. Previous studies investigating readability and suitability of PEMs utilize various tools, including the SAM Tool, the suitability and comprehensibility assessment instrument (SAM + CAM), and the PEMAT, to name a few. Likewise, this study pulled from multiple studies to determine the best method for analyzing online KD resources. Whereas other studies used multiple search engines (e.g., Google, Bing) to create the resource list, this study used only one search engine.

Discussion of a genetic condition inherently includes complex terminology. Readability scores rely on the number of syllables in words and sentences. Even when defined, complex terms increase the readability score. This study did not include “pop-up definitions” in the analysis. Some resources used genetics jargon in their main text and used a pop-up capability to display definitions when the user moused over the word. This information was not captured in the readability formulas because text was simply copied and pasted from the main page.

Furthermore, there are limitations to the analysis tools used in this study. The SAM Tool is inherently subjective. Multiple reviewers analyzed each resource using this tool to achieve some level of objectivity. Even with multiple reviewers, scores may be discrepant due to different interpretations of factors. The SAM Tool is also limited in its use for online resources, as it was

originally designed for physical copies of PEMs (Doak et al., 1985). Typography factors, such as paper quality, are not appropriate for website analysis. The PEMAT Audio/Visual version can be used for multimedia materials like videos, audio tracks/podcasts, and interactive graphics. According to the PEMAT user guide, the tool should be used in conjunction with other suitability tools because the PEMAT does not include readability in its assessment of understandability. Content criteria was determined by the authors from clinical and family perspectives. Inclusion of these criteria for KD resources may be subject to affinity bias. Quality or accuracy of content was not investigated in this study.

2.4.2 Future Research

Results and conclusions from this study support those of similar previous studies – PEMs are written well above the recommended reading levels and are not entirely suitable for target audiences. Future researchers should use these results and recommendations to develop updated online resources for KD. The current study was conducted as a descriptive review of currently available online KD resources. It would be beneficial to include parents, a key stakeholder, in the conversation. Researchers could expand on this study by exploring parent perception of PEMs, current resources and resources improved based on this study's recommendations. Theoretically, the recommendations in this study will improve current resources; however, the impact for patients and their families is unknown unless directly studied. This study also did not analyze accuracy of information in online KD resources. Learning about patient's knowledge base prior to genetic counseling appointments and how they use that information to make informed decisions can allow counselors to tailor sessions to meet patients' personal needs.

2.5 Conclusions

Health literacy infiltrates every aspect of healthcare and has been declared by Healthy People 2030 to be a foundational principle and overarching public health goal. Low health literacy is a core public health concern due to its impact on health outcomes. Accessible and understandable resources are one key component to increasing patient comprehension and empowerment, as well as improving health outcomes. This study highlighted the mediocre quality of current online KD patient education materials. Overall, resources struggled using common language to make the information understandable to the broader population, incorporating interactivity and presenting concrete next steps based on information presented, and utilizing graphics appropriately. This lack of the use of appropriate reading level and other components leading to low suitability has the potential to interact with low health literacy in the population and result in misunderstanding and negative health outcomes.

3.0 Project Significance to Genetic Counseling and Public Health

Rare diseases, like KD, used to be considered of insignificant public health impact. Today, however, the combined impact of rare diseases that collectively affect millions of individuals worldwide has been recognized as a public health genetics issue. Approximately 10 percent of Americans are living with a rare disease, 80 percent of which are genetic in origin (Plaiasu, Nanu, & Matei, 2010). Without appropriate, understandable PEMs, patients may be left with a dearth of information, and health disparities may be amplified. To date, online KD resources have not been analyzed for readability and suitability. This study reinforced the conclusions of prior research studies that showed PEMs continue to grapple with readability and suitability. After analyzing online KD resources, it is easy to understand the importance of PEMs being accessible to the general population. This research serves as the basis for future research to ensure PEMs are readable and appropriate for parents whose child is diagnosed with KD.

Public health professionals are trained to protect and promote the health of individuals and communities, considering the contribution of social determinants of health (SDOH) to health disparities and inequities. “SDOH are the conditions in the environments where people are born, live, learn, work, play, worship, and age that affect a wide range of health, functioning, and quality-of-life outcomes and risks” (HealthyPeople2030). SDOH include economic stability, education access and quality, healthcare access and quality, neighborhood and built environment, and social and community context. A general framework and measurable goals for progression toward ensuring equitable access to quality healthcare were created by various federal agencies, including the Centers for Disease Control and Prevention (CDC) (“10 Essential Public Health Services,”

2023). This framework, known as the 10 Essential Public Health Services, extends to all public health initiatives including PEMs and, specifically, online KD education materials.

This study encompasses two of the 10 Essential Public Health Services: 1) communicate effectively to inform and educate, and 2) improve and innovate through evaluation, research, and quality improvement. Low health literacy is an indirect health hazard, leading to worse health outcomes due to medication noncompliance and insufficient understanding prior to providing consent, to name a few (Shahid et al., 2022). Without superior PEMs, people with low health literacy are left to flounder. Health literacy is currently considered a “research objective” by Healthy People 2030. As such, we must invest resources into examining and updating research on health literacy and ways to aid in individuals’ understanding before health disparities can then be improved. The research status of this public health issue may qualify it for future research grants to improve currently existing PEMs and to expand beyond this study of resources for KD to PEMs for other rare diseases.

As a sub entity of the larger public health genetics network, genetic counseling is also affected by this research. Genetic counselors have training in handling potentially traumatic and/or anxiety-provoking situations and work to convey the necessary, often complicated, information in a sensitive manner. They play a role in creating and referring to supplemental educational materials and locating support resources for parents (Marcus, 2019). Peterson and colleagues found, during the NBS process, some parents were given limited information by non-genetics providers after their child received a diagnosis of KD (Peterson, Siemon, Olewiler, McBride, & Allain, 2022).

Not only are genetic counselors educating patients, but they also serve as consultants for other healthcare providers. Doctors, primary care physicians, and nurse practitioners are often the

referral pipeline to genetic counselors. Improved online KD resources can bolster providers' genetics knowledge pertaining to KD, resulting in improved referrals. Non-genetics providers may also benefit from this research to build their library of patient education resources.

While genetic counselors are hand-picking which resources to share with patients and colleagues, they are not regularly running systematic analyses on the resources. It is imperative that counselors are aware of the readability, suitability, and accessibility of patient resources to allow for more targeted, personal resource referrals. Furthermore, although genetic counselors have specialized training to explain complex topics in layman's terms, these providers are typically not present when patients access online resources, making it even more important that these materials are explicit and suitable. Presenting genetic information at a lower reading level where possible or being diligent to explain complex jargon is a simple solution to consider when creating patient resources. Suitable and accessible online KD resources may also aid in diminishing the genetics knowledge gap for providers. As many providers recognize their lack of genetics expertise, they refer to genetic counselors for accurate risk assessment and information (French et al., 2023).

The conclusions from this study are relevant to public health and genetic counseling because they establish the need for updated online resources for parents whose children are diagnosed with rare diseases, specifically KD. With this data and similar findings in previous research, we can take steps forward to communicate effectively and drive innovation through evaluation and quality improvement. This study is the groundwork for developing improved KD resources, as future researchers build upon the Healthy People 2030 framework to increase health literacy.

Appendix A Thesis IRB Exemption



Ries, John William
To: Zieber, Megan Elizabeth



Mon 10/9/2023 11:30 AM

Hey Megan,

Thank you again for taking the time to meet with me today to discuss your upcoming Thesis work. Based on the nature of your research provided in a previous email, as well as what we discussed over our call, it is clear that your thesis work does not require IRB oversight. This is due to the fact that your research is considered 'Not Human Subjects Research' as per the federal regulations. This determination is based on the following conditions:

- This research study does not involve human subjects as:
 - All data will be de-identified in accordance with HIPAA.
 - No identifiable private information will be reviewed or recorded by the study team.
 - No member of the research team has interacted or intervened, for research purposes, with the individuals whose data.

Best of luck with your research and please reach out with any additional questions as they arise.

Take care,
John

Appendix B List of Resources by Google Search Term

Krabbe disease

- <https://www.ninds.nih.gov/health-information/disorders/krabbe-disease>
- <https://medlineplus.gov/genetics/condition/krabbe-disease/>
- <https://www.chp.edu/our-services/rare-disease-therapy/conditions-we-treat/krabbe-disease>
- <https://my.clevelandclinic.org/health/diseases/6039-krabbe-disease-globoid-cell-leukodystrophy>
- <https://www.huntershope.org/family-care/leukodystrophies/krabbe-disease/>
- <https://www.mountsinai.org/health-library/diseases-conditions/krabbe-disease>
- <https://rarediseases.org/rare-diseases/leukodystrophy-krabbes/>
- https://en.wikipedia.org/wiki/Krabbe_disease
- https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=GB&Expert=487
- <https://www.childrenshospital.org/conditions/krabbe-disease>

Globoid cell leukodystrophy

- <https://medlineplus.gov/genetics/condition/krabbe-disease/>
- <https://www.ninds.nih.gov/health-information/disorders/krabbe-disease>
- <https://my.clevelandclinic.org/health/diseases/6039-krabbe-disease-globoid-cell-leukodystrophy>
- <https://vcahospitals.com/know-your-pet/globoid-cell-leukodystrophy-in-dogs>

- <https://www.chp.edu/our-services/rare-disease-therapy/conditions-we-treat/krabbe-disease>
- <https://rarediseases.org/rare-diseases/leukodystrophy-krabbes/>
- <https://www.huntershope.org/family-care/leukodystrophies/krabbe-disease/>
- <https://www.childrenshospital.org/conditions/krabbe-disease>
- https://en.wikipedia.org/wiki/Krabbe_disease
- <https://jamanetwork.com/journals/jamaneurology/article-abstract/569694>

Parent resources for Krabbe disease

- <https://krabbeconnect.org/krabbe-disease/patient-and-caregivers/krabbe-disease-patient-support-team/>
- <https://www.chp.edu/our-services/rare-disease-therapy/conditions-we-treat/krabbe-disease>
- <https://krabbeconnect.org/krabbe-disease/patient-and-caregivers/resources/>
- <https://www.huntershope.org/family-care/leukodystrophies/krabbe-disease/>
- <https://krabbefacts.org/>
- <https://www.childneurologyfoundation.org/disorder/krabbe-disease/>
- <https://www.mountsinai.org/health-library/diseases-conditions/krabbe-disease>
- <https://rarediseases.org/organizations/krabbeconnect/>
- <https://www.babysfirsttest.org/newborn-screening/conditions/krabbe>
- <https://www.merckmanuals.com/home/children-s-health-issues/hereditary-metabolic-disorders/krabbe-disease>

GALC deficiency

- https://www.chp.edu/our-services/rare-disease-therapy/locations?utm_mrid=mrid2319&utm_source=GOOGLE&utm_medium=cpc&utm_campaign=71700000089012583&utm_adgroup=58700007513803417&utm_term=kra bbe+disease&utm_advertiserid=700000002154619&gclid=EAIAIQobChMIx4Tx-6iwwgMVTllHAR1f7AaWEAAYASAAEgL-svD_BwE&gclsrc=aw.ds
- <https://www.mountsinai.org/health-library/diseases-conditions/krabbe-disease>
- <https://pubmed.ncbi.nlm.nih.gov/36113749/>
- <https://www.ncbi.nlm.nih.gov/books/NBK1238/>
- [https://www.orpha.net/consor/cgi-bin/Disease_Search.php?lng=EN&data_id=22&Disease_Disease_Search_diseaseType=ORPHA&Disease_Disease_Search_diseaseGroup=487&Disease\(s\)/group%20of%20diseases=GALC-deficiency&title=GALC-deficiency&search=Disease_Search_Simple](https://www.orpha.net/consor/cgi-bin/Disease_Search.php?lng=EN&data_id=22&Disease_Disease_Search_diseaseType=ORPHA&Disease_Disease_Search_diseaseGroup=487&Disease(s)/group%20of%20diseases=GALC-deficiency&title=GALC-deficiency&search=Disease_Search_Simple)
- <https://medlineplus.gov/genetics/gene/galc/>
- <https://rarediseases.org/rare-diseases/leukodystrophy-krabbes/>
- <https://www.sciencedirect.com/science/article/pii/S0969996122002546>
- <https://www.uptodate.com/contents/krabbe-disease/print>
- <https://www.babysfirsttest.org/newborn-screening/conditions/krabbe>

Krabbe disease newborn screening

- https://www.chp.edu/our-services/rare-disease-therapy/locations?utm_mrid=mrid2319&utm_source=GOOGLE&utm_medium=cpc&utm_campaign=71700000089012583&utm_adgroup=58700007513803417&utm_term=kra

bbe+disease&utm_advertiserid=700000002154619&gclid=EAIaIQobChMI0a26yKqwgwMVs0hHAR0j1grmEAAAYASAAEgIf5PD_BwE&gclsrc=aw.ds

- https://www.bio-rad.com/en-us/a/cd/newborn-screening?WT.mc_id=231003039374&WT.srch=1&WT.knsh_id=51da2e67-b566-4f54-8e05-d91c18b747e9&s_kwcid=AL!18120!3!676893719650!!!g!!&gad_source=1&gclid=EAIaIQobChMI0a26yKqwgwMVs0hHAR0j1grmEAAAYAiAAEgK4NvD_BwE
- <https://www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritable-disorders/krabbe-27-june-2018.pdf>
- <https://www.babysfirsttest.org/newborn-screening/conditions/krabbe>
- <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4014301/>
- <https://newbornscreening.hrsa.gov/conditions/krabbe-disease>
- <https://everylifefoundation.org/newborn-screening-take-action/krabbe-disease-kd/>
- <http://www.idph.state.il.us/HealthWellness/fs/krabbe.htm>
- <https://ojrd.biomedcentral.com/articles/10.1186/s13023-018-0766-x>
- <https://dph.illinois.gov/content/dam/soi/en/web/idph/files/publications/krabbe-idph-fact-sheet-121317.pdf>

Appendix C List of Resources Analyzed

Organizations are listed in a random order to maintain anonymity. This order does not correspond to the order of Organizations A-L in the manuscript.

Baby's First Test¹

HRSA²

MedlinePlus³

Hunter's Hope⁴

EveryLife Foundation⁵

KrabbeConnect⁶

NORD⁷

United Leukodystrophy Foundation⁸

KrabbeFacts⁹

The Jackson Project¹⁰

Child Neurology Foundation¹¹

Genetic and Rare Diseases Information Center¹²

1. (Baby's First Test, 2024) 2. (HRSA, 2023) 3. (MedlinePlus, 2018) 4. (Hunter's Hope, 2024) 5. (EveryLife Foundation, 2023) 6. (KrabbeConnect, 2024) 7. (NORD, 2020) 8. (United Leukodystrophy Foundation, 2024) 9. (KrabbeFacts, 2024) 10. (The Jackson Project, 2014) 11. (Child Neurology Foundation, 2024) 12. (Genetic and Rare Diseases Information Center, 2023)

Appendix D Readability and Suitability Scores

Table 8 Resource Readability Scores

Resource	Flesch-Kincaid Grade Level	SMOG Reading Grade Level
Organization A	11.8	13.5
Organization B	12.8	14.6
Organization C	10.5	12.7
Organization D	10.6	12.5
Organization E	10.1	12.1
Organization F	7.4	9.8
Organization G	8.0	10.5
Organization H	10.6	12.4
Organization I	12.4	12.7
Organization J	10.0	12.0
Organization K	10.3	12.4
Organization L	12.9	14.2

Table 9 Average SAM Tool Scores and Interpretations

Resource	Average SAM Tool Score (%)	Score Interpretation
Organization A	53.4	Adequate
Organization B	54.9	Adequate
Organization C	46.6	Adequate
Organization D	57.0	Adequate
Organization E	80.7	Superior
Organization F	60.3	Adequate
Organization G	46.5	Adequate
Organization H	42.8	Adequate
Organization I	48.9	Adequate
Organization J	68.6	Adequate
Organization K	38.4	Not suitable
Organization L	43.1	Adequate
TOTAL Average	53.4	---

Table 10 Average PEMAT Scores and Interpretations

Resource	Average Understandability Score (%)	Score Interpretation	Average Actionability Score (%)	Score Interpretation
Organization A	77.08	Adequate	24.44	Not suitable
Organization B	61.54	Not suitable	0	Not suitable
Organization C	69.23	Not suitable	0	Not suitable
Organization D	74.36	Adequate	60.00	Adequate
Organization E	90.63	Adequate	83.33	Adequate
Organization F	75.00	Adequate	0	Not suitable
Organization G	69.23	Not suitable	60.00	Not suitable
Organization H	55.05	Not suitable	0	Not suitable
Organization I	62.02	Not suitable	0	Not suitable
Organization J	94.12	Adequate	41.67	Not suitable
Organization K	55.88	Not suitable	41.67	Not suitable
Organization L	53.85	Not suitable	10.00	Not suitable
TOTAL Averages	69.83	---	26.76	---

Appendix E SAM Tool Ratings

Table 11 SAM Ratings Part 1

Reviewer	Organization A		Organization B		Organization C	
	A	B	A	B	A	B
Purpose is Evident	1	1	2	1	1	1
Content about Behaviors	0	1	0	0	0	0
Scope is Limited	2	2	2	2	2	2
Summary or Review Included	0	1	0	0	0	0
Reading Grade Level	0	0	0	0	0	0
Writing Style, Active Voice	1	1	1	0	0	1
Vocabulary Uses Common Words	1	0	2	1	1	1
Context is Given First	1	1	2	1	1	1
Learning Aids via “Road Signs”, Subtitles and Captions	2	2	2	2	2	2
Cover Graphic Shows Purpose	1	1	2	2	2	2
Type of Graphics	2	1	2	2	0	0
Relevance of Illustrations	2	2	2	1	0	0
Lists and Tables Explained	0	0	n/a	0	0	0
Captions Used for Graphics	0	0	0	0	0	0
Layout Factors	2	2	2	1	1	1
Typography	2	2	2	2	2	2
Subheads (“chunking”) Used	2	1	1	0	2	2
Interaction Used	0	1	0	0	0	1
Behaviors are Modeled and Specific	1	1	1	1	1	0
Motivation, Self-Efficacy	0	1	1	1	1	2
Match in Logic, Language, Experience (LLE)	2	2	2	2	2	2
Cultural Image and Examples	1	1	1	1	1	2
TOTAL	23	24	27	20	19	22
# N/A	0	0	1	0	0	0
#N/A x 2	0	0	2	0	0	0
44 – (N/A) x 2	44	44	42	44	44	44
(Revised Max Score)						
TOTAL / Revised Max	0.523	0.545	0.643	0.455	0.432	0.500
Interpretation	Adequate	Adequate	Adequate	Adequate	Adequate	Adequate

Table 12 SAM Ratings Part 2

Reviewer	Organization D		Organization E		Organization F	
	A	B	A	B	A	B
Purpose is Evident	1	1	2	2	1	1
Content about Behaviors	1	1	1	2	0	0
Scope is Limited	2	2	2	2	2	2
Summary or Review Included	1	1	2	1	2	2
Reading Grade Level	0	0	0	0	0	0
Writing Style, Active Voice	1	2	1	2	1	2
Vocabulary Uses Common Words	2	1	1	1	2	1
Context is Given First	2	1	2	2	2	1
Learning Aids via “Road Signs”, Subtitles and Captions	2	2	2	2	2	2
Cover Graphic Shows Purpose	1	1	2	2	2	1
Type of Graphics	n/a	0	2	2	1	1
Relevance of Illustrations	0	0	2	2	1	1
Lists and Tables Explained	1	1	1	2	0	0
Captions Used for Graphics	0	0	0	0	0	0
Layout Factors	2	2	2	2	2	2
Typography	2	2	2	2	2	2
Subheads (“chunking”) Used	1	1	2	2	2	2
Interaction Used	0	0	1	1	0	0
Behaviors are Modeled and Specific	1	1	2	2	1	1
Motivation, Self-Efficacy	2	2	2	2	1	2
Match in Logic, Language, Experience (LLE)	2	2	2	2	1	2
Cultural Image and Examples	1	1	1	2	1	2
TOTAL	25	24	34	37	26	27
# N/A	1	0	0	0	0	0
#N/A x 2	2	0	0	0	0	0
44 – (N/A) x 2 (Revised Max Score)	42	44	44	44	44	44
TOTAL / Revised Max	0.595	0.545	0.773	0.841	0.591	0.614
Interpretation	Adequate	Adequate	Superior	Superior	Adequate	Adequate

Table 13 SAM Ratings Part 3

Reviewer	Organization G		Organization H		
	A	B	A	B	C
Purpose is Evident	0	1	1	1	2
Content about Behaviors	0	0	0	0	1
Scope is Limited	2	2	1	2	2
Summary or Review Included	0	1	0	1	0
Reading Grade Level	0	0	0	0	0
Writing Style, Active Voice	1	2	0	1	1
Vocabulary Uses Common Words	1	1	1	1	0
Context is Given First	1	1	1	1	0
Learning Aids via “Road Signs”, Subtitles and Captions	2	2	2	2	0
Cover Graphic Shows Purpose	0	0	1	1	1
Type of Graphics	n/a	0	1	0	1
Relevance of Illustrations	0	0	1	0	1
Lists and Tables Explained	0	1	n/a	0	n/a
Captions Used for Graphics	0	0	n/a	0	n/a
Layout Factors	2	2	1	1	2
Typography	2	2	2	2	2
Subheads (“chunking”) Used	2	1	0	0	2
Interaction Used	1	0	0	1	0
Behaviors are Modeled and Specific	1	1	0	0	1
Motivation, Self-Efficacy	1	1	0	1	2
Match in Logic, Language, Experience (LLE)	2	2	2	2	1
Cultural Image and Examples	1	1	1	1	1
TOTAL	19	21	15	18	20
# N/A	1	0	2	0	2
#N/A x 2	2	0	4	0	4
44 – (N/A) x 2 (Revised Max Score)	42	44	40	44	40
TOTAL / Revised Max	0.452	0.477	0.375	0.409	0.500
Interpretation	Adequate	Adequate	Not suitable	Adequate	Adequate

Table 14 SAM Ratings Part 4

	Organization I		Organization J		Organization K	
	A	B	A	B	A	B
Reviewer						
Purpose is Evident	1	1	2	1	0	1
Content about Behaviors	0	0	0	1	0	0
Scope is Limited	2	2	2	2	1	1
Summary or Review Included	0	0	2	2	0	1
Reading Grade Level	0	0	0	0	0	0
Writing Style, Active Voice	0	1	1	2	0	0
Vocabulary Uses Common Words	0	0	1	2	0	0
Context is Given First	1	0	1	1	1	1
Learning Aids via “Road Signs”, Subtitles and Captions	2	2	2	2	2	2
Cover Graphic Shows Purpose	1	1	1	0	1	1
Type of Graphics	1	1	2	1	n/a	0
Relevance of Illustrations	1	1	2	2	0	0
Lists and Tables Explained	n/a	1	n/a	1	1	1
Captions Used for Graphics	2	2	2	1	0	0
Layout Factors	2	2	2	2	1	1
Typography	2	2	2	2	2	2
Subheads (“chunking”) Used	2	1	2	1	2	1
Interaction Used	0	0	0	0	0	0
Behaviors are Modeled and Specific	0	0	1	1	1	1
Motivation, Self-Efficacy	n/a	1	1	2	1	1
Match in Logic, Language, Experience (LLE)	2	2	2	2	2	2
Cultural Image and Examples	1	1	1	2	1	1
TOTAL	20	21	29	30	16	17
# N/A	2	0	1	0	1	0
#N/A x 2	4	0	2	0	2	0
44 – (N/A) x 2 (Revised Max Score)	40	44	42	44	42	44
TOTAL / Revised Max	0.500	0.477	0.690	0.682	0.381	0.386
Interpretation	Adequate	Adequate	Adequate	Adequate	Not suitable	Not suitable

Table 15 SAM Ratings Part 5

Reviewer	Organization L	
	A	B
Purpose is Evident	1	1
Content about Behaviors	0	0
Scope is Limited	1	1
Summary or Review Included	0	1
Reading Grade Level	0	0
Writing Style, Active Voice	1	1
Vocabulary Uses Common Words	1	1
Context is Given First	1	1
Learning Aids via “Road Signs”, Subtitles and Captions	2	2
Cover Graphic Shows Purpose	1	1
Type of Graphics	0	0
Relevance of Illustrations	0	0
Lists and Tables Explained	n/a	0
Captions Used for Graphics	0	0
Layout Factors	1	2
Typography	2	2
Subheads (“chunking”) Used	2	1
Interaction Used	0	0
Behaviors are Modeled and Specific	1	1
Motivation, Self-Efficacy	1	1
Match in Logic, Language, Experience (LLE)	2	2
Cultural Image and Examples	1	1
TOTAL	18	19
# N/A	1	0
#N/A x 2	2	0
44 – (N/A) x 2	42	44
(Revised Max Score)		
TOTAL / Revised Max	0.429	0.432
Interpretation	Adequate	Adequate

Appendix F PEMAT Ratings

Table 16 PEMAT Understandability Scores Part 1

Reviewer	Organization A			Organization B	
	A	B	C	A	B
Purpose is Completely Evident	0	1	1	1	0
Does not include information that distracts from purpose	1	1	1	1	1
Uses Common, Everyday Language	0	0	1	0	0
Medical terms used only to familiarize audience	1	0	1	1	0
Uses Active Voice	1	0	1	0	1
Numbers are clear and easy to understand	1	1	1	1	1
User not expected to perform calculations	1	1	1	1	1
Material is broken into “chunks”/short sections	1	1	1	1	0
Sections have informative headers	1	1	1	1	1
Information is presented in logical sequence	1	1	1	1	1
Summary Provided	0	0	0	0	0
Visual cues used to draw attention to key points	1	1	1	1	1
Visual aids used to make content easier to understand	1	1	1	0	0
Visual aids reinforce rather than distract from content	1	1	1	n/a	n/a
Visual aids have clear titles or captions	0	0	0	n/a	n/a
Illustrations are clear and uncluttered	1	1	1	n/a	n/a
Tables are simple with short, clear headings	n/a	n/a	n/a	n/a	n/a
TOTAL Points	12	11	14	9	7
TOTAL Possible Points	16	16	16	13	13
Understandability score (%)	75.00	68.75	87.50	69.23	53.85
Interpretation	Adequate	Not suitable	Adequate	Not suitable	Not suitable

Table 17 PEMAT Understandability Scores Part 2

Reviewer	Organization C		Organization D			Organization E	
	A	B	A	B	C	A	B
Purpose is Completely Evident	0	1	0	1	1	1	1
Does not include information that distracts from purpose	1	1	1	1	1	1	1
Uses Common, Everyday Language	1	1	1	0	0	1	1
Medical terms used only to familiarize audience	0	0	1	1	1	1	1
Uses Active Voice	1	0	0	1	1	1	1
Numbers are clear and easy to understand	1	1	1	1	1	1	1
User not expected to perform calculations	1	1	1	1	1	1	1
Material is broken into “chunks”/short sections	1	1	1	1	1	1	1
Sections have informative headers	1	1	1	1	1	1	1
Information is presented in logical sequence	1	1	0	1	1	1	1
Summary Provided	0	0	0	1	0	1	1
Visual cues used to draw attention to key points	1	1	1	1	1	1	1
Visual aids used to make content easier to understand	0	0	0	0	0	1	1
Visual aids reinforce rather than distract from content	n/a	n/a	n/a	n/a	n/a	1	1
Visual aids have clear titles or captions	n/a	n/a	n/a	n/a	n/a	0	0
Illustrations are clear and uncluttered	n/a	n/a	n/a	n/a	n/a	1	0
Tables are simple with short, clear headings	n/a	n/a	n/a	n/a	n/a	n/a	n/a
TOTAL Points	9	9	8	11	10	15	14
TOTAL Possible Points	13	13	13	13	13	16	16
Understandability score (%)	69.23	69.23	61.54	84.62	76.92	93.75	87.50
Interpretation	Not suitable	Not suitable	Not suitable	Adequate	Adequate	Adequate	Adequate

Table 18 PEMAT Understandability Scores Part 3

Reviewer	Organization F			Organization G		Organization H	
	A	B	C	A	B	A	B
Purpose is Completely Evident	1	1	1	0	0	0	1
Does not include information that distracts from purpose	1	1	1	1	1	1	1
Uses Common, Everyday Language	1	1	1	1	0	1	0
Medical terms used only to familiarize audience	1	1	1	1	1	0	0
Uses Active Voice	0	1	1	0	1	0	0
Numbers are clear and easy to understand	1	1	1	1	1	1	1
User not expected to perform calculations	1	1	1	1	1	1	1
Material is broken into “chunks”/short sections	1	1	1	1	1	1	1
Sections have informative headers	1	1	1	1	1	1	1
Information is presented in logical sequence	1	1	1	1	1	1	1
Summary Provided	1	1	0	0	0	0	0
Visual cues used to draw attention to key points	0	1	1	1	1	0	0
Visual aids used to make content easier to understand	0	0	0	0	0	0	0
Visual aids reinforce rather than distract from content	0	0	0	n/a	n/a	1	n/a
Visual aids have clear titles or captions	0	0	0	n/a	n/a	0	n/a
Illustrations are clear and uncluttered	1	1	1	n/a	n/a	1	n/a
Tables are simple with short, clear headings	n/a	n/a	n/a	n/a	n/a	n/a	n/a
TOTAL Points	11	13	12	9	9	9	7
TOTAL Possible Points	16	16	16	13	13	16	13
Understandability score (%)	68.75	81.25	75.00	69.23	69.23	56.25	53.85
Interpretation	Not suitable	Adequate	Adequate	Not suitable	Not suitable	Not suitable	Not suitable

Table 19 PEMAT Understandability Scores Part 4

Reviewer	Organization I		Organization J		Organization K	
	A	B	A	B	A	B
Purpose is Completely Evident	0	0	1	1	0	1
Does not include information that distracts from purpose	1	1	1	1	1	0
Uses Common, Everyday Language	0	0	1	1	0	0
Medical terms used only to familiarize audience	0	0	1	1	0	0
Uses Active Voice	1	1	0	0	0	1
Numbers are clear and easy to understand	1	1	1	1	1	1
User not expected to perform calculations	1	1	1	1	1	1
Material is broken into “chunks”/short sections	1	1	1	1	1	1
Sections have informative headers	1	1	1	1	1	1
Information is presented in logical sequence	1	1	1	1	1	1
Summary Provided	0	0	1	1	0	0
Visual cues used to draw attention to key points	0	1	1	1	1	1
Visual aids used to make content easier to understand	0	0	1	1	0	1
Visual aids reinforce rather than distract from content	1	n/a	1	1	1	0
Visual aids have clear titles or captions	1	n/a	1	1	0	0
Illustrations are clear and uncluttered	1	n/a	1	1	1	0
Tables are simple with short, clear headings	n/a	n/a	1	1	1	0
TOTAL Points	10	8	16	16	10	9
TOTAL Possible Points	16	13	17	17	17	17
Understandability score (%)	62.50	61.54	94.12	94.12	58.82	52.94
Interpretation	Not suitable	Not suitable	Adequate	Adequate	Not suitable	Not suitable

Table 20 PEMAT Understandability Scores Part 5

Reviewer	Organization L	
	A	B
Purpose is Completely Evident	0	0
Does not include information that distracts from purpose	0	0
Uses Common, Everyday Language	0	0
Medical terms used only to familiarize audience	1	0
Uses Active Voice	1	1
Numbers are clear and easy to understand	1	1
User not expected to perform calculations	1	1
Material is broken into “chunks”/short sections	1	1
Sections have informative headers	1	1
Information is presented in logical sequence	1	1
Summary Provided	1	0
Visual cues used to draw attention to key points	0	0
Visual aids used to make content easier to understand	0	0
Visual aids reinforce rather than distract from content	n/a	n/a
Visual aids have clear titles or captions	n/a	n/a
Illustrations are clear and uncluttered	n/a	n/a
Tables are simple with short, clear headings	n/a	n/a
TOTAL Points	8	6
TOTAL Possible Points	13	13
Understandability score (%)	61.54	46.15
Interpretation	Not suitable	Not suitable

Table 21 PEMAT Actionability Scores Part 1

Reviewer	Organization A			Organization B		Organization C	
	A	B	C	A	B	A	B
Clearly identifies at least one action the user can take	1	0	1	0	0	0	0
Addresses the user directly when describing actions	0	0	0	0	0	0	0
Breaks down any action into manageable, explicit steps	0	0	0	0	0	0	0
Provides a tangible tool whenever it could help the user take action	0	0	0	0	0	0	0
Provides simple instructions or examples of how to perform calculations	n/a	n/a	n/a	n/a	n/a	n/a	n/a
Explains how to use charts, graphs, tables, or diagrams to take actions	0	n/a	n/a	n/a	n/a	n/a	n/a
Uses visual aids whenever they could make it easier to act on instructions	1	1	0	0	0	0	0
TOTAL Points	2	1	1	0	0	0	0
TOTAL Possible Points	6	5	5	5	5	5	5
Understandability score (%)	33.33	20.00	20.00	0	0	0	0
Interpretation	Not suitable	Not suitable	Not suitable	Not suitable	Not suitable	Not suitable	Not suitable

Table 22 PEMAT Actionability Scores Part 2

Reviewer	Organization D			Organization E	
	A	B	C	A	B
Clearly identifies at least one action the user can take	1	1	1	1	1
Addresses the user directly when describing actions	1	1	1	1	1
Breaks down any action into manageable, explicit steps	1	1	0	1	1
Provides a tangible tool whenever it could help the user take action	1	0	0	1	1
Provides simple instructions or examples of how to perform calculations	n/a	n/a	n/a	n/a	n/a
Explains how to use charts, graphs, tables, or diagrams to take actions	n/a	n/a	n/a	0	0
Uses visual aids whenever they could make it easier to act on instructions	0	0	0	1	1
TOTAL Points	4	3	2	5	5
TOTAL Possible Points	5	5	5	6	6
Understandability score (%)	80.00	60.00	40.00	83.33	83.33
Interpretation	Adequate	Not suitable	Not suitable	Adequate	Adequate

Table 23 PEMAT Actionability Scores Part 3

Reviewer	Organization F			Organization G		Organization H	
	A	B	C	A	B	A	B
Clearly identifies at least one action the user can take	0	0	0	1	1	0	0
Addresses the user directly when describing actions	0	0	0	1	1	0	0
Breaks down any action into manageable, explicit steps	0	0	0	0	1	0	0
Provides a tangible tool whenever it could help the user take action	0	0	0	1	0	0	0
Provides simple instructions or examples of how to perform calculations	n/a	n/a	n/a	n/a	n/a	n/a	n/a
Explains how to use charts, graphs, tables, or diagrams to take actions	n/a	n/a	n/a	n/a	n/a	n/a	n/a
Uses visual aids whenever they could make it easier to act on instructions	0	0	0	0	0	0	0
TOTAL Points	0	0	0	3	3	0	0
TOTAL Possible Points	5	5	5	5	5	5	5
Understandability score (%)	0	0	0	60.00	60.00	0	0
Interpretation	Not suitable	Not suitable	Not suitable	Not suitable	Not suitable	Not suitable	Not suitable

Table 24 PEMAT Actionability Scores Part 4

Reviewer	Organization I		Organization J		Organization K	
	A	B	A	B	A	B
Clearly identifies at least one action the user can take	0	0	1	1	1	1
Addresses the user directly when describing actions	0	0	1	1	1	1
Breaks down any action into manageable, explicit steps	0	0	0	0	0	0
Provides a tangible tool whenever it could help the user take action	0	0	1	0	0	0
Provides simple instructions or examples of how to perform calculations	n/a	n/a	n/a	n/a	n/a	n/a
Explains how to use charts, graphs, tables, or diagrams to take actions	n/a	n/a	0	0	0	0
Uses visual aids whenever they could make it easier to act on instructions	0	0	0	0	1	0
TOTAL Points	0	0	3	2	3	2
TOTAL Possible Points	5	5	6	6	6	6
Understandability score (%)	0	0	50.00	33.33	50.00	33.33
Interpretation	Not suitable	Not suitable	Not suitable	Not suitable	Not suitable	Not suitable

Table 25 PEMAT Actionability Scores Part 5

Reviewer	Organization L	
	A	B
Clearly identifies at least one action the user can take	1	0
Addresses the user directly when describing actions	0	0
Breaks down any action into manageable, explicit steps	0	0
Provides a tangible tool whenever it could help the user take action	0	0
Provides simple instructions or examples of how to perform calculations	n/a	n/a
Explains how to use charts, graphs, tables, or diagrams to take actions	n/a	n/a
Uses visual aids whenever they could make it easier to act on instructions	0	0
TOTAL Points	1	0
TOTAL Possible Points	5	5
Understandability score (%)	20.00	0
Interpretation	Not suitable	Not suitable

Table 26 PEMAT Audio/Visual Ratings

Reviewer	Organization B	
	A	B
Purpose is Completely Evident	1	1
Uses Common, Everyday Language	1	1
Medical terms used only to familiarize audience	1	1
Uses Active Voice	0	1
Material is broken into “chunks”/short sections	0	1
Sections have informative headers	0	0
Information is presented in logical sequence	1	1
Summary Provided	1	1
Visual cues used to draw attention to key points	1	1
Text on screen is easy to read	1	1
Allows user to hear the words clearly	1	1
Illustrations are clear and uncluttered	1	1
Tables are simple with short, clear headings	n/a	1
TOTAL Points	9	12
TOTAL Possible Points	12	13
Understandability score (%)	75.00	92.31
Interpretation	Adequate	Adequate
Clearly identifies at least one action the user can take	0	0
Addresses the user directly when describing actions	0	0
Breaks down any action into manageable, explicit steps	0	0
Explains how to use charts, graphs, tables, or diagrams to take actions	n/a	0
TOTAL Points	0	0
TOTAL Possible Points	3	4
Actionability score (%)	0	0
Interpretation	Not suitable	Not suitable

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