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Mapping the evidence of multidimensional health approaches in treating individuals with Prader–Willi Syndrome: A scoping review protocol

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ABSTRACT

Introduction Prader–Willi Syndrome (PWS) is one of the rare diseases involving genetics and affects various body systems. The disease is known due to the absence of paternal genes on chromosome 15q11-q13. Multisystem complex conditions require interdisciplinary healthcare treatment. However, to the best of our knowledge, there is little evidence of an established successful model of an interdisciplinary approach in managing rare diseases like PWS.

Methods and analysis The scoping review process follows the five-staged Arksey and O'Malley (2005) methodology framework excluding the optional consultation stage (stage 6): the definition of the research questions (step 1); the eligibility criteria and search strategy are defined (stage 2); the study selection process based on the eligibility criteria identified will follow (stage 3); a framework developed for this review will then inform the extraction and charting of data from the included studies (step 4) and results will be aggregated and summarised with criteria relevant for health professionals and policymakers (stage 5). We will search for electronic databases (MEDLINE/PubMed, Scopus, Web of Science), grey literature sources and critical studies' reference lists to determine the appropriate inclusion criteria. Three researchers will review all abstracts and full-text studies for inclusion.

Ethics and dissemination This scoping review methodology does not require ethical approval since it aims to synthesise information from available publications. A scoping review article will be submitted for publication to a scientific journal following this protocol.

Strengths and limitations of this study

- This protocol is the plan for the future scoping review work to deal with the extensive literature on multidimensional approaches in treating Prader–Willi Syndrome (PWS) individuals.
- MEDLINE/PubMed, Scopus and Web of Science electronic databases and grey literature sources will be utilised to retrieve relevant articles associated with the topic of discussion as many as possible.
- Articles published only in the English language will be included in the scoping review.
- Articles from grey literature will be critically evaluated and appraised to determine the quality of the information using six criteria from the AACODS checklist.
- The review will cover children, adolescents and adults with PWS and issues with the interdisciplinary transitioning care over the life span will be addressed.

BACKGROUND

Prader–Willi Syndrome (PWS) is a rare genetic disease involving a multisystem of the affected body. The syndrome is due to the absence of paternal genes on chromosome 15q11-q13. PWS has characteristic phenotypes, including physical, cognitive, mental and behavioural symptoms, with numerous implications for metabolic, endocrine and neurologic systems.[1 2] The syndrome also shows changing clinical features during a patient's life[3] . Commonly, complex multisystem disorders require interdisciplinary healthcare treatment. However, to the best of our knowledge, there is little evidence of an established successful model of a multidisciplinary approach in managing rare diseases like PWS. Thus, a synthesis of current evidence is needed to inform researchers and relevant health professionals on the interdisciplinary management models suitable for PWS. As of this date, the model for multidisciplinary approaches for PWS existed in France and the Netherlands[4] . Duis *et al*[5] proposed a Centre of Excellence utilising an interdisciplinary, patient-centred and family-centred approach. The purpose of this scoping review protocol is to map out the evidence of a multidimensional approach in delivering comprehensive care by collating services from interdisciplinary specialists.

This scoping review protocol is presented to inform the future scoping review article. The methodology involves reviewing the extensive body of literature through mega electronic

databases and grey literature resources to get an overall picture of this topic, summarise the outcomes from the published studies, and thus, identifying the research gaps[6] . The scoping review's aim is not to critically appraise individual studies that may be heterogeneous in terms of their research design, methodology and differences in the quality of outcomes reported[7] . This review protocol will focus on several available previous scoping reviews on different treatments of PWS individuals and adopt a comprehensive interdisciplinary treatment approach. Second, there is a limited systematic review on multidimensional therapy approaches for PWS over the last 10 years.[5 8] This scoping review protocol was conducted to map specific clinical services for PWS and the parents/caregivers and family's roles in providing care.

METHOD AND ANALYSIS

Patient and public involvement

No patient is involved in this scoping review protocol.

Conceptual model

Since the original description of PWS in 1956, this complex disorder continues to unfold as one of the most complex syndromes in genetics, physiology and behaviour, making it the most challenging syndrome to manage. The multidimensional problems with PWS require an interdisciplinary team approach for holistic patient care.[9] Implementing a multidisciplinary treatment approach that includes diagnosis and rehabilitation professionals should result in better outcomes, and subsequently, reduce healthcare expenses.[10 11] There were suggestions that the clinic staff should consist of specialists in paediatric neurology, child psychiatry, endocrinology, psychology, orthopaedics and nutrition. Consultations with respiratory physicians and Ear, Nose and Throat surgeons depend on the patient's needs.[9 12] The medical treatment's primary aims are to normalise weight for height.[13 14] Comprehensive medical care includes growth hormone therapy for children with a documented growth hormone/1GF axis deficiency and short stature. Dietary management comprises planned weight management programmes that include dietary supervision, calorie restrictions, nutritional planning and education for parents and caregivers. The physical therapy approach is for hypotonia, and psychosocial therapy involves behavioural and cognitive interventions and psychotropic intervention.[15]

Protocol design

The scoping review follows the framework proposed by Arksey and O'Malley[7] and developed further by Levac *et al*[16] and the Joanna Briggs Institute.[17] The review process involves at least five stages: stage 1—identifying the research question; stage 2—identifying relevant studies; stage 3—study selection; stage 4—charting the data and stage 5—collating, summarising and reporting the results. The optional stage 6 from the framework proposed by Arksey and O'Malley[7] will be excluded. Stage 6 involves a consultation exercise with a panel of experts for possible extra references to be included and get their feedback on the outcomes of the scoping review. Even though it is useful to consult with a panel of experts in stage 6, we are constrained by time and budget factors. The detailed protocol for each step is described below.

Stage 1: identifying the research questions

First, the available literature on multi/interdisciplinary treatment approaches for PWS individuals will be explored to refine the literature's present scope and identify the research questions. In this phase, no criteria will be outlined to avoid the review's restriction to any study populations as treatment should start as early as possible.

Based on the initial exploratory research, the following research questions were identified:

1. What are the biopsychosocial profiles or clinical characteristics of individuals with PWS?
2. Which measures are used to assess multiple problems in individuals with PWS?
3. Are there any specific interdisciplinary treatment modules developed for individuals with PWS?
4. What is the main focus of treatment for individuals with PWS in the literature?
5. In what setting are these interventions provided?
6. Are there any treatment modules involving parents/caregivers and family members?
7. Are these interventions effective?
8. How do interventions/treatment modules and their assessments change over the lifespan of the individual with PWS?
9. What are the scales used to measure the effectiveness of the treatment modules?
10. What are the factors involved in preventing or facilitating effective implementation of the interventions?
11. What are the indicators to prove the effectiveness of the interventions, either alone or in combination?
12. Are the currently used assessments of interdisciplinary treatment approaches adequate to capture the impact and benefits of treatment?

Stage 2: identifying relevant studies

The second stage aims to identify the inclusion criteria for the scoping review, which will help the researchers find the relevant resources from the broad spectrum of the literature.

Articles selected for the scoping review include published original papers and systematic reviews, which can be accessed through several electronic databases: MEDLINE/PubMed, Scopus and Web of Science. Grey literature sources include Google Scholar, government's reports, clinical practice guidelines and published or unpublished thesis. Articles from grey literature will be critically evaluated and appraised to determine the quality of the information using six criteria from the AACODS checklist (*Authority, Accuracy, Coverage, Objectivity, Date, Significance*) developed by Jessica Tyndall from the Flinders University Medical Library.[18] Retrieved

articles from the electronic resources will be scrutinised to fit in with the inclusion criteria.

Following stage 1, the inclusion criteria are set as below:

- Publication types: journal articles and grey literature sources.
- Time frame: 10 years (2010–2020).
- Language: English.
- Study population: children to adults with PWS.
- Types of intervention: specific intervention or a combination of treatment approaches for a particular problem or multiproblems of individuals with PWS.
- Types of review articles: narrative reviews, scoping reviews, systematic reviews, meta-analyses.

Following the first stage of scoping process, we listed the exclusion criteria as follows: abstracts of conferences, proceedings, magazines, book reviews, commentaries or editorial articles and reviews focusing on interventions for the treatment of comorbidity in PWS (eg, Autism Spectrum Disorder or Attention Deficit Hyperactivity Disorder or psychiatric comorbidities).

Based on a suggestion by Levac *et al*,[16] an iterative process will be utilised to identify the key search terms: PWS, treatment, multidimensional approach, multi/interdisciplinary approach and review. The selected review articles will undergo screening for the titles, abstracts and index search terms. A consultation with a librarian is vital to get advice on the most suitable Medical Subject Headings Terms (MESH terms) for the search and change them according to the different databases. This approach will ensure extensive coverage of the literature. Following the initial exploratory scoping phase, the search strings for each database will be listed. Finally, articles from the different databases will be compiled and imported into reference manager software.

Stage 3: study selection

This stage involves the process of identifying the relevant literature to be included in the scoping review. The retrieved articles were compiled from different databases, and duplicate articles will be removed. As mentioned earlier, articles from grey literature, including sources such as Google Scholar, will be critically evaluated and appraised to determine the quality of the information using six criteria from the AACODS checklist.[18] Articles that do not meet the inclusion criteria set in stage 2 will be excluded. Finally, the full paper of those eligible following the titles and abstracts' screening will be retrieved.

Another team member will screen a sample (ie, 20%) of the retrieved articles to ensure consistency in following the review's inclusion criteria. The second person will also review any titles and abstracts that the first reviewer is unsure of including if any. Any disagreements concerning the eligibility of the sampled articles will be solved through discussion between the three reviewers/authors or the appointment of a fourth reviewer if required. The Preferred Reporting Items for Systematic Reviews and Meta-Analyses flowchart will be used to summarise the study selection process, which will be updated once the review is complete.

Stage 4: charting the data

Following the initial exploratory review, a data extraction framework consisting of 18 categories was developed. These categories will be used to assess the eligibility criteria of the full-review articles retrieved (see table 1). The review's type and objectives will be reported alongside standard bibliographical information (ie, authors, title, journal and year of publication) for published journal articles and grey literature sources. In addition, for each piece, information covered by the reviews on biopsychosocial profiles of PWS, assessments, setting, types of intervention, duration and intensity of the interventions, the outcomes to be assessed, factors influencing the effectiveness of the interventions, factors that facilitate and prevent successful implementation of the interventions will be tabled.

Two team members will do pilot-testing on a sample of at least 10% of the completed list of retrieved studies to ensure the consistency of the coding framework. Modification of the categories and revision of the data extraction framework will be done accordingly, if necessary. The team will discuss questions when piloting the framework and possible disagreements through team consultations. All team members will chart the data from selected studies independently and guided by the data extraction framework. A sample (ie, 20%) of the selected articles independently reviewed will then be compared among the team members to control for inter-rater reliability. Any discrepancies that occur during this data extraction process will be sorted out among the three reviewers until a consensus is reached or through a fourth reviewer if required.

Stage 5: collating, summarising and reporting the results

This stage involves data analysis of eligible articles using the 18-category data extraction framework developed in stage 4. The results of these analyses should respond to the 12 research questions outlined in stage 1. In addition, the data should provide information on essential issues studied on multidimensional, interdisciplinary interventions for PWS individuals, children to adults, which include transition care. For example, a study by Duis *et al*[5] on a multidisciplinary approach to clinical management of PWS children proposed an interdisciplinary clinic that supports persons with complex genetic diseases and their families through integrated care. The scoping review study will also highlight the different treatment and clinical effectiveness of the interventions reviewed (eg, single approach or interdisciplinary approaches) and their impact on the particular problem or multiple problems in the PWS population. Stage 5 also highlights areas that have been less explored or research gaps and provides opportunities for future research. Results from the data analysis will be summarised collectively using tables and charts, whichever is appropriate.

Table 1 Data extraction framework

Main category	Subcategory	Description
1. Authors		
2. Title		
3. Journal		
4. Year of publication		
5. Objectives of the review		Outline the objectives of the scoping study
6. Type of review		State the type of review of the article selected, such as narrative, scoping, systematic, meta-analysis or other types of review
7. Number of studies included in the review		Indicate the number of primary studies included in the review
8. Years of publication of the studies included in the review		Specify the range of the years of publications of the studies included in the review
9. Type of studies included in the review		State the specific types of studies included for the review (eg, case study, case–control, cross-sectional, random control trials (RCTs), etc)
10. Characteristics of the intervention(s)	► Intervention types ► Method of intervention delivery ► Duration and intensity of the intervention	► State the type(s) of the interventions ► The description of how the intervention was done and who provided the intervention ► State the duration of intervention and its frequency
11. Biopsychosocial profiles of PWS children, adolescents and adults		Describe the strengths and weaknesses of PWS individuals in their biological profiles, physical functions, behavioural characteristics, nutritional status and cognitive profiles
12. Multidimensional measurements of functions		Describe instruments commonly used to screen or measure multidimensional functioning in PWS children to adults
13. The setting of the intervention(s)		State whether the interventions were performed in school-based, family-based or community-based settings
14. Outcomes of the intervention studies		Reported outcomes of the interventions done in the review (eg, weight, height, BMI, behaviour, cognitive function)
15. Effectiveness of the intervention studies		Describe the changes occur in the reviewed studies as measures of the effectiveness of the intervention (eg, change in cognitive status, quality of life)
16. Impact on the study population		Describe the impact on the study population through the quality of life measure post-intervention
17. Factors supporting intervention		Report on the factors that influence or facilitate the implementation of the interventions in the study reviewed
18. Factors against intervention		Report on the factors that prevent the implementation of the interventions reported in the review

BMI, body mass index; PWS, Prader–Willi Syndrome.

Ethics and dissemination

This scoping review study does not require ethical approval since the methodology involves synthesising information from publicly available publications. Dissemination of outcomes is through an article write-up for a journal publication and a conference presentation.

The results from the scoping review will provide essential information to policymakers, the PWS associations and health professionals interested in planning, funding and delivering evidence-based and effective PWS individuals' interventions on children, adolescents and adults care and highlight gaps or missing evidence. The information will also be shared with other professionals involved in the interdisciplinary treatment of PWS individuals across the life span in future workshops. For example, evidence on integrated clinical management of PWS individuals applying professionals from various disciplines[5] and integrating the experiences caring and living of PWS persons and their families through research and cohesive treatment modalities.[19] It will also show appropriate timelines to intervene, and suitable intervention approaches to ensure effectiveness as the transition in clinical features according to age and treatment is imperative in PWS. It will also highlight the most appropriate interventions to ensure clinical effectiveness (eg, medication trials for hyperphagia and food-related behaviours in PWS). The scoping review will also inform us of the impact of the intervention on the population in need (eg, the effect of transitional care on endocrine and anthropometric parameters in PWS) and will explore the adequacy of assessment to capture the impact and benefit of interdisciplinary treatment approaches.

Collaborators This protocol paper is an initial work from our multidisciplinary research study involving Prader-Willi Syndrome individuals and their families. This research project is a collaboration between the Faculty of Health Sciences, Universiti Kebangsaan Malaysia (UKM) and the Prader-Willi Syndrome Association (PWSA) Malaysia.

Contributors All authors have made substantial intellectual contributions to the development of this protocol. NCD conceptualised the review approach, initiated the first draft of the manuscript. NCD, CU, NHAW and ABT conceived the scoping review and developed the research questions and the search strategy. All authors critically reviewed the revised drafts, edited the manuscript and approved the final version of the manuscript.

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Competing interests None declared.

Patient and public involvement Patients and/or the public were not involved in the design, or conduct, or reporting or dissemination plans of this research.

Patient consent for publication Not required.

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