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A participatory mixed-method study protocol to develop, implement and evaluate a Care Management model for patients with Neuromuscular Diseases (Care-NMD-CH Study)

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Abstract

Background: Neuromuscular diseases (NMD) are progressive, leading to reduced quality of life and life expectancy, while requiring long-term caregiving. As patients age, caregivers face increasing demands, often resulting in significant caregiver burden. Treatment and care are complex, and a coordinated, family-centred approach is indicated. The current care situation for individuals with NMD and their families in Switzerland is poorly understood, and there is no standard for NMD care management available.

Objective: The proposed study aims to assess the current care practices for individuals with NMD in Switzerland, identifying unmet needs and challenges faced by patients, families, and health care providers. Based on these findings, a care management model will be developed, implemented, and evaluated to enhance support structures, strengthen specialist health centres, and improve overall health care outcomes for affected individuals and their families.

Methods: We planned a three-phase participatory mixed-methods study design. First, qualitative descriptive and quantitative survey data will be used to determine the current state of care practices. Then, a care management model and training program are co-developed based on these insights. Last, the care management service is implemented and evaluated in pilot specialist health centers. The protocol is published after the completion of data collection ("Data Existing" IRRID) and is intended to serve as guidance for similar projects in the future.

Results: Expected outcomes include enhanced patient and caregiver satisfaction, improved quality of life and self-efficacy, reduced caregiver burden and hospitalizations, better inter-professional collaboration, and reduced healthcare costs through more coordinated, family-centred care.

Conclusions: Evidence-based and family-centred care management is expected to strengthen the support system for patients living with these rare but severe conditions and to create a sustainable care situation despite limited financial resources in the future. Expected outcomes include improved quality of life, self-efficacy and family functioning and reduced caregiver burden and health care costs.

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A participatory mixed-method study protocol to develop, implement and evaluate a Care Management model for patients with Neuromuscular Diseases (Care-NMD-CH Study)

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Abstract

Background

Neuromuscular diseases (NMD) are progressive, leading to reduced quality of life and life expectancy, while requiring long-term caregiving. As patients age, caregivers face increasing demands, often resulting in significant caregiver burden. Treatment and care are complex, and a coordinated, family-centred approach is indicated. The current care situation for individuals with NMD and their families in Switzerland is poorly understood, and there is no standard for NMD care management available.

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The proposed study aims to assess the current care practices for individuals with NMD in Switzerland, identifying unmet needs and challenges faced by patients, families, and health care providers. Based on these findings, a care management model will be developed, implemented, and evaluated to enhance support structures, strengthen specialist health centres, and improve overall health care outcomes for affected individuals and their families.

Methods

We planned a three-phase participatory mixed-methods study design. First, qualitative descriptive and quantitative survey data will be used to determine the current state of care practices. Then, a care management model and training program are co-developed based on these insights. Last, the care management service is implemented and evaluated in pilot specialist health centers. The protocol is published after the completion of data collection (“Data Existing” IRRID) and is intended to serve as framework for similar projects in the future.

Results

Expected outcomes include enhanced patient and caregiver satisfaction, improved quality of life and self-efficacy, reduced caregiver burden and hospitalizations, better inter-professional collaboration, and reduced healthcare costs through more coordinated, family-centred care.

Conclusions

Evidence-based and family-centred care management is expected to strengthen the support system for patients living with these rare but severe conditions and to create a sustainable care situation despite limited financial resources in the future. Expected outcomes include improved quality of life, self-efficacy and family functioning and reduced caregiver burden and health care costs.

Keywords: Neuromuscular Diseases, Rare Diseases, Care Management, Caregivers, Quality of Life, Caregiver Burden, Mixed Methods, Study Protocol

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Introduction

Many neuromuscular diseases (NMD) follow a degenerative course. They can be associated with a high burden of disease, a reduced quality of life and a shortened life expectancy [1,2]. However, thanks to new disease-modifying therapies, improvements in symptomatic treatment, and advanced health technologies, NMD now often have a better prognosis than ever before [3-5]. Despite these advancements, individuals with NMD still require regular therapies as well as assistance in activities of daily living. In many families with affected children or adults, family members take over caregiving responsibilities [6-8]. As individuals with NMD grow older, their caregivers must continue providing care for longer periods, which poses additional practical and psychosocial challenges [9]. The prolonged and intensive caregiving often leads to an increased caregiver burden [7,10,11].

Treatment and care of NMD patients is inherently complex and requires a coordinated, multidisciplinary, and interprofessional approach [12-16]. Families benefit from targeted support, assistance with care, care coordination, and family-strengthening interventions [6,8,17]. This is particularly relevant during health crises and important stages such as the loss of ambulation, the affected young person's transition into adulthood, and the end-of-life phase [7,18]. It is strongly advised to use a systemic approach when caring for the well-being of the individual with NMD and the entire family.

Strengthening the support system for individuals living with these rare but often serious diseases is essential to ensure a sustainable care model for the future. The current care situation for individuals with NMD in Switzerland, however, is poorly understood. There is no standard for NMD care management available. Care management services provided by health centres can support care coordination, improve the quality of life of frequent health care users, and enhance health care provider satisfaction, all while potentially reducing costs to the health care system [19,20]. Care management prioritizes interdisciplinary collaboration and effective communication among all the professionals involved in patient care to plan, implement, coordinate, evaluate, and prioritize services based on patient needs [21]. Through care management, individuals living with NMD are empowered, and their self-efficacy and ability to self-manage their condition can improve [20,22,23].

There is a need for an evidence-based, coordinated and family centred approach to care for the NMD population [24,25] to strengthen the support system for patients living with these rare and often severe conditions, and to create a sustainable care situation despite limited financial resources in the future.

Study Purpose and Objectives

The purpose of this study is to:

a) describe the current state of care practices for individuals living with NMD within a specific cohort and identify unmet needs and challenges related to patients, their families and the care team;
b) develop a care management model and training program capable of addressing identified gaps in the support chain and enhancing the support offered by specialist health centres and
c) implement the care management model and evaluate its impact for patients with NMD and their families, for the interprofessional care team and for the health care system.

Methods

Study design

The study uses a participatory mixed methods study design that includes qualitative and quantitative data collection at different stages [26]. This design is suitable for studies that address health disparities through empowerment of marginalized or underrepresented populations, involve the target population in the research [26] and examine complex phenomena, such as the evaluation of services where various perspectives need to be included [27,28]. The study will use a pragmatic philosophical stance and a family systems theoretical framework [29]. Data integration occurs through methods (building, merging) and during interpretation (joint display, narrative) [26].

Setting and participants

Initial steps involve identifying the study cohort, establishing an expert panel (physicians, advanced practice nurses and researchers) and sounding board (affected individuals, family members, professionals and other stakeholder) for feedback, guidance and quality control, and creating a participant recruitment plan. The study cohort of interest for this study (Care-NMD-CH study) are NMD patients and their families living in Switzerland. In Switzerland, seven neuromuscular centres provide specialized outpatient and inpatient care for patients with NMD. These centers are hospital-based tertiary national reference centres. Four groups of study participants were identified: 1) patients with NMD; 2) family members (parents, siblings, partner and other relatives); 3) professionals who are involved in the care and other stakeholders (e.g. nurses, physiotherapists, members of patient organisations). Participants include members of all Swiss language regions and ages. Recruitment includes the identification of collaborating institutions and gatekeepers, who inform potential participants about study participation.

Study phases

According to the study aims, there are three phases A, B, C over a duration of five years (2020-2025). In Phase A, qualitative and quantitative data are collected with instruments tailored to the needs of the individuals being studied to assess the current state of care practice [30]. In Phase B, a new care management model and training program is developed involving relevant stakeholders, including patients with NMD, their families, and health care professionals. In Phase C, the new care management model is implemented in pilot specialist health centres and evaluated using a pre-test and post-test design. This project was funded in 2020, the care management service was

implemented in March 2023, and data collection was completed in March 2025. At the time of submission, data analysis is ongoing. The protocol is therefore published after the completion of data collection (“Data Existing” IRRID) and is intended to serve as guidance for similar projects in the future. The following criteria are used to reflect on the quality of the study: rigor, credibility, and transferability [31-33].

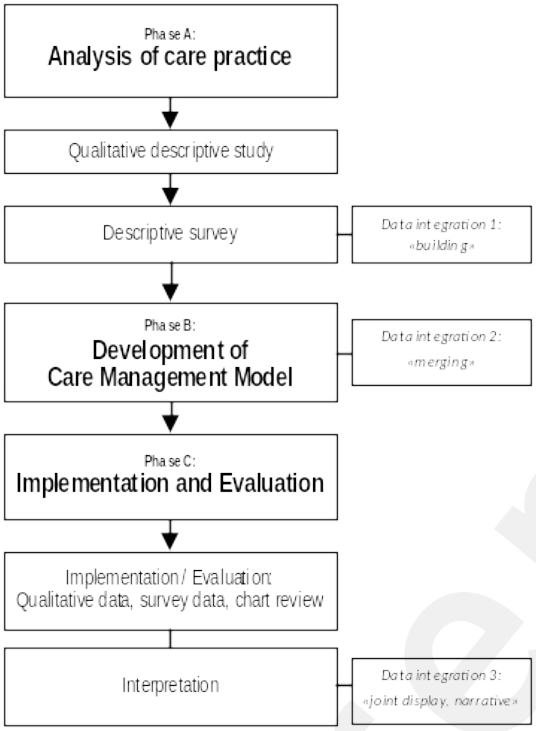

Figure 1: Overview of the study phases

Study Phase A: Analysis of the current state of care practice

1. Qualitative descriptive study

At first, a qualitative-descriptive research design is applied to explore and describe the current state of care practice [34,35]. This approach facilitates close engagement with the data and enables a subjective exploration of people's everyday experiences and needs [36,37]. A purposive sampling strategy will be employed, which selects respondents that will yield appropriate and helpful information [38]. Data are collected through one-on-one and focus group interviews with patients with NMD, family members, professionals who are involved in the care and other stakeholders (n= 45, patients and family members 8 years and older).

Qualitative data collection and analysis

Separate semi-structured interview guides are developed for each participant group containing open-ended and probing questions. Questions assess the general care situation, benefits of care, gaps in the support chain, unmet needs, experiences within the care network, barriers to sustainability, communication and collaboration. Audio data are transcribed verbatim according to the guidelines set

by Dresing and Pehl [39]. The latest version of MAXQDA is used for data analysis [40].The transcripts are analysed using qualitative data analysis principles outlined by Saldaña [41].The inductive analysis process contains four steps: 1) open coding without using prior concepts or predefined categories; 2) code allocation into main areas; 3) bundling into subcategories; and 4) comparison and revision of subcategories and generation of categories. The socio-demographic data of participants are collected and analysed descriptively.

2. Descriptive survey

The quantitative part of phase A consists of an online descriptive survey to collect further information on the current practices of care from a larger group of participants. The results from the qualitative study are used to develop the survey components. This is the first data integration point in our mixed methods study, where results from one data collection (qualitative) inform the next data collection (quantitative), an integration called “building” [26,30].

Quantitative data collection

The online survey includes self-developed questions, based on the obtained qualitative results and standardized instruments (Table 1). Four questionnaires are developed, one for each group of participants (patients, family members, professionals, and other stakeholders; n=500, patients and family members aged 14 years and older). The questionnaires consist of Likert-type questions, Visual Analogue Scale, closed and open-ended questions. Topics common to all groups of addressed participants include socio-demographics, information about the care situation, satisfaction with care, gaps in the support network/services, unmet needs, experiences within the care network, barriers to sustainability, and collaboration. Group-specific topics include quality of life, self-efficacy for patients, family functioning and caregiver burden for family members, quality of care, and interprofessional collaboration for professionals. The survey is conducted and managed using web-based REDCap electronic data capture tools [42]. The survey is made available online, and participants are recruited using a multi-tiered approach to gather a range of viewpoints. To ensure anonymization of data and to be able to link data over different time points, each participant marks the questionnaire with a personal code developed according to a defined scheme.

Table 1: Overview of survey components

	Patients with NMD	Family members	Professionals	Other stakeholder
Component 1	Self- developed questions based on the qualitative study			
Component 2	Health-related quality of life, WHO Quality of Life Questionnaire (WHOQOL-BREF) [43]	Family Functioning; Family Assessment Device (FAD) [44]	Quality of Care, quality of care single-item measure [45]	Acute events/hospitalisation, morbidity, mortality (Documents / Statistics)
Component 3	Self-efficacy (General Self-Efficacy Scale, GSE) [46]	Caregiver Burden; Burden Scale for Family Caregivers (BSFC – short version) [47]	Interprofessional collaboration (IPC) [48]	Costs / Financing / Reimbursement (Documents / Statistics)

Quantitative data analysis

The dataset is examined for missing values and out-of-range values using frequency tables. This is reported if a variable under study has ≤5% missing values. If more than 5% of the values in the total score of an instrument are missing, they are imputed using the Expectation-Maximization (EM) algorithm in EM regression estimation, if necessary and feasible [49]. The data analysis is carried out using the latest version of IBM SPSS. We describe the sample using frequencies, percentiles, means, standard deviations, and ranges. The instrument's internal consistency, measured via Cronbach's alpha, must be above 0.80. Minimum values of 0.60 can be accepted for small sample sizes and scales with few items. The significance level is set at <0.05. Quantitative findings are discussed with the expert panel and sounding board to check for clinical relevance and credibility.

Study Phase B: Development of a Care Management Model

The care management model is developed following the structure of a logic model [50]. Therefore, the results from the qualitative and quantitative phase A are integrated to delineate unmet needs and challenges for the study groups, which can be addressed by care managers. This is the second data integration point in our mixed methods study, where integration of data occurs for comparison through “merging” quantitative and qualitative findings from Phase A [26,30]. The developed model outlines the roles and responsibilities of NMD care managers and contains a comprehensive requirement and competency profile that covers both pediatric and adult care. Experts from the expert panel and sounding board are involved in the development of the model following a creative cyclical process of repeated phases of reflection, planning, action, observation, reflection, and re-planning [51].

Based on the developed care management model, a training program for NMD care managers is developed, which consists of classroom and online learning sessions, and covers a broad spectrum of essential topics related to NMD-specific patient care. Key areas include: pathophysiology, diagnostics, supportive interventions, as well as nursing and treatment options; psychosocial aspects, palliative care, and the relevant transitions (e.g., from pediatric to adult care); systemic counselling, interprofessional collaboration, and leadership. Care managers are expected to network, complete an internship, and prepare a case report. The two-year part-time training program starts with the implementation of the care management in the pilot specialist health centres.

Study Phase C: Implementation and evaluation

Implementation

Phase C comprises the recruitment of pilot specialist health centres for the two-year implementation and a context analysis ahead of the implementation. The context analysis is necessary to better understand the specific institutional situation, to overcome implementation challenges and to identify factors that may influence intervention uptake and study outcomes [52,53]. The context analysis is based on the Integrated Promoting Action on Research Implementation in Health Services (I-PARISH) Framework [52,54]. The analysis is performed through focus group interviews (n=8) using a semi-structured interview guide based on the I-PARISH

implementation framework, which focuses on recipients, evidence, inner (local) and outer (organization) context [54]. The aim is to identify barriers and facilitators to adapt the NMD care management model to individual institutional needs. Interviews are audio recorded and transcribed verbatim and analyzed as outlined in Phase A.

Evaluation

The implementation process and outcome of the care management will be evaluated using formative and summative evaluation strategies. Formative evaluation assesses the local implementation strategy, monitors the process, and allows for real-time adjustments, improving the chances of success [55,56]. This is achieved through the training program, regular contact between the clinical team and research team, and documentation of clinical practice. After each contact with patients and their families or any other activity related to their role, this information is documented by the care manager and collected by the research team for further analysis (care manager record). The record includes anonymized data, such as the type of contact, counselling topics, assessment, and quality of life. Summative evaluation of the care management model is done by collecting qualitative and quantitative data through the care manager records, interviews and surveys on four levels and at different time points (T1-T4) (Table 2).

Table 2: Overview data collection across time points

Time	Phase A	Phase B	Phase C				
Participants	**T0, Baseline**	**Development of Care Management Model**	**Context analysis**	**Implementation start**	**T1, 6 months**	**T2, 12 months**	**T3, 18 months**	**T4, 24 months**
Patients with NMD	Int, S		-			S, R	Int	S, R
Family members	Int, S		-		-	S	Int	S
Professionals	Int, S		Int		Int	S	-	S
Stakeholders	Int, S		-		-	S	-	S

Note: Int: Interviews, S: Survey, R: Care Manager Record

Qualitative Descriptive Study, data collection and analysis

Six months after the implementation start (T1), focus group interviews are conducted with professionals involved in the care at the pilot specialist health centres to evaluate the implementation and impact of the NMD care management. Following 18 months after implementation (T3), semi-structured interviews are undertaken with patients and their families to assess the potential benefits of the care management services. The semi-structured interview guides, data transcription, and analysis correspond to those applied for the qualitative study of Phase A.

Quantitative data collection and analysis

The survey described in Phase A is repeated in Phase C under the same conditions: The same quantitative data are collected at 12 months (T2) and 24 months (T4) after the implementation start.

Data integration during interpretation

Finally, findings will be integrated through “joint displays” and “narrative” at a third data integration point [26]. Therefore, both qualitative and quantitative findings are reported together on a theme-by-theme basis for selected themes (weaving approach). Quantitative data will present the general trends and patterns, while qualitative data will be used to illustrate and deepen understanding of those trends through participant narratives. This integrated approach will be used in reporting and presentations to ensure a coherent and holistic account of the study's outcomes. Additionally, data are brought together through a joint display using a table or matrix to draw out new insights beyond the information gained separately [26]. Therefore, a triangulation protocol will be applied.

Ethical considerations

This study adheres to Swiss national laws and the ethical principles of the Declaration of Helsinki [57]. Approval was received from the relevant cantonal ethics committee (BASEC ID 2020-01882). Informed consent of the participants will be obtained. All participants will be informed that their participation is voluntary and that they can withdraw from the study at any time. For data security, consent forms, and other documents containing identifying information will be stored in a restricted archive for 10 years after the study ends.

Expected outcomes of the study

The proposed study investigates the current care situation for individuals with NMD and their families in Switzerland. It aims to develop, implement, and evaluate a care management targeted at this population. The study aims to achieve holistic improvements in clinical outcomes, patient and family well-being, and health care service efficiency. Expected outcomes include enhanced patient and caregiver satisfaction through more coordinated, family-centred care. Developing a care management model with patients, family members, and professionals is expected to result in a comprehensive model. This model will meet real-life needs that have the potential to improve significantly health-related quality of life, self-efficacy, and family functioning, while simultaneously reducing caregiver burden and the frequency of acute health care events such as hospitalizations. Improved collaboration among health care professionals within the specialist centres and with external institutions through information exchange and optimized resource utilization, thus potentially reducing overall health care costs and creating a more sustainable model for NMD care. Additionally, the conduction of this study aims to build and strengthen a network of specialized care managers for NMD within and across health care settings, facilitate the education and exchange of NMD expertise, establish a standard for care management in NMD, and develop a training program for NMD care managers. This participatory mixed-methods study protocol developed for the Care-NMD-CH study can be adapted and applied to other settings and patient populations with complex health care needs, providing a scalable and generalizable framework for improving care management practices.

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Authors' contributions

VW, HP, GMS, MW, BGM, RW and MK contributed to the study concept. CS and VW drafted the manuscript. All authors critically revised the content of the manuscript and read and approved the final manuscript.

The authors used ChatGPT (Open AI) for language editing and proofreading during the final phase of this manuscript. We confirm that ChatGPT or any other artificial intelligence generated tools were not used to develop this research project or the content in this manuscript. The authors take full responsibility for the content and interpretation of the manuscript.

Competing interests

GMS served on advisory boards of AveXis, Biogen, Novartis, Pfizer, Roche and Italfarmaco, received speaker’s honoraria from Novartis, and is a member of the steering board of the Swiss registry for neuromuscular disorders (Swiss-Reg-NMD). All other authors declare that they have no competing interests.

Availability of data and material

The data collection instruments and templates are available from the corresponding author on reasonable request.

Abbreviations

I-PARISH	Integrated Promoting Action on Research Implementation in Health Services
NMD	Neuromuscular Disease

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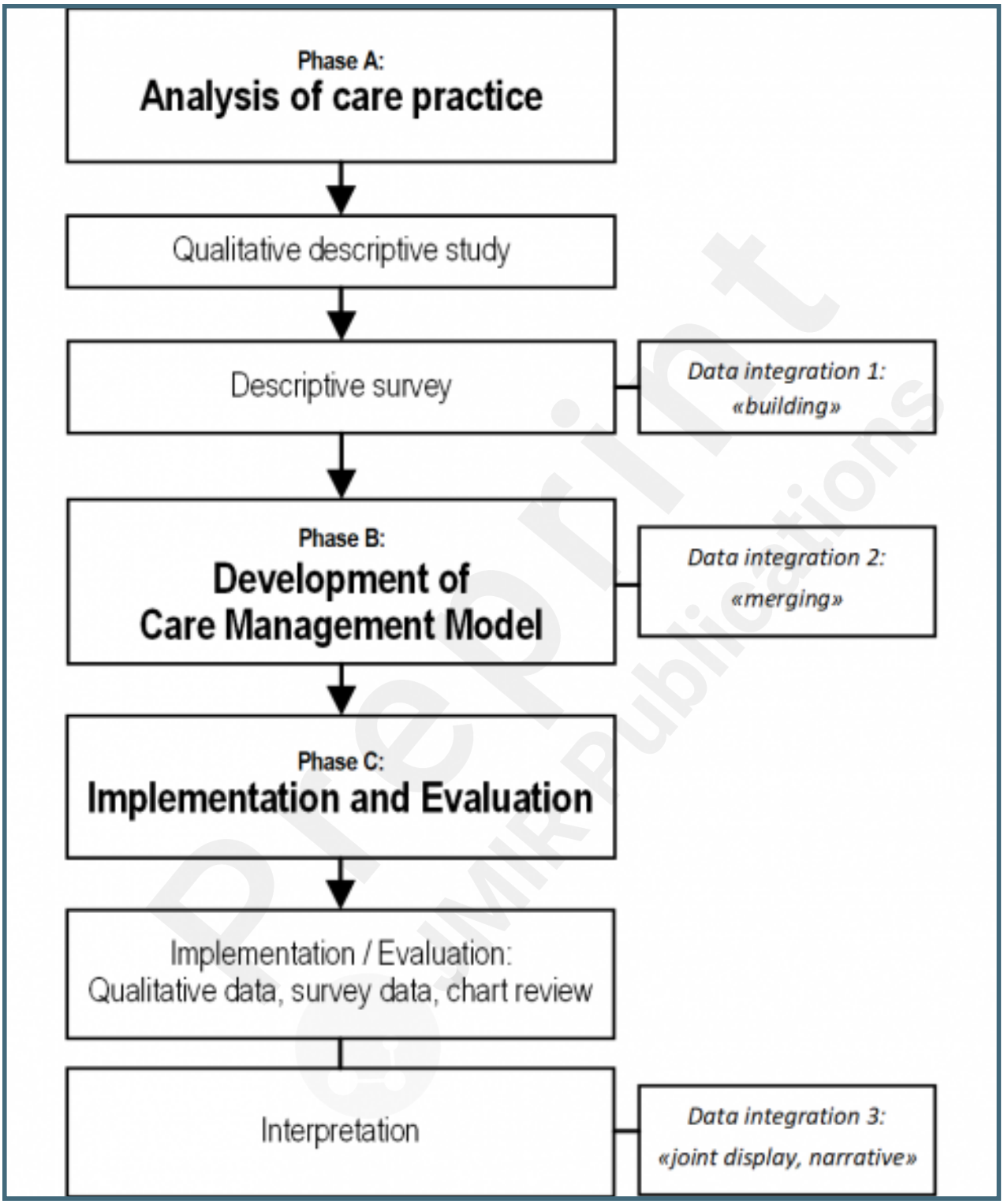
Overview of the study phases.



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