Developing a taxonomy of care coordination for people living with rare conditions: a qualitative study

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Abstract

Background: Improving care coordination is particularly important for individuals with rare conditions (who may experience multiple inputs into their care, across different providers and settings). To develop and evaluate strategies to potentially improve care coordination, it is necessary to develop a method for organising different ways of coordinating care for rare conditions. Developing a taxonomy would help to describe different ways of coordinating care and in turn facilitate development and evaluation of pre-existing and new models of care coordination for rare conditions. To the authors' knowledge, no studies have previously developed taxonomies of care coordination for rare conditions. This research aimed to develop and refine a care coordination taxonomy for people with rare conditions.

Methods: This study had a qualitative design and was conducted in the United Kingdom. To develop a taxonomy, six stages of taxonomy development were followed. We conducted interviews (n = 30 health care professionals/charity representatives/commissioners) and focus groups (n = 4 focus groups, 22 patients/carers with rare/ultra-rare/undiagnosed conditions). Interviews and focus groups were audio-recorded with consent, and professionally transcribed. Findings were analysed using thematic analysis. Themes were used to develop a taxonomy, and to identify which types of coordination may work best in which situations. To refine the taxonomy, we conducted two workshops (n = 12 patients and carers group; n = 15 professional stakeholder group).

Results: Our taxonomy has six domains, each with different options. The six domains are: (1) Ways of organising care (local, hybrid, national), (2) Ways of organising those involved in care (collaboration between many or all individuals, collaboration between some individuals, a lack of collaborative approach), (3) Responsibility for coordination (administrative support, formal roles and responsibilities, supportive roles and no responsibility), (4) How often appointments and coordination take place (regular, on demand, hybrid), (5) Access to records (full or filtered access), and (6) Mode of care coordination (face-to-face, digital, telephone).

Conclusions: Findings indicate that there are different ways of coordinating care across the six domains outlined in our taxonomy. This may help to facilitate the development and evaluation of existing and new models of care coordination for people living with rare conditions.

Keywords: Rare conditions, Rare diseases, Care coordination, Taxonomy, Qualitative, Health care organisation

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Introduction

The complexity of the organisation and delivery of health care has been further complicated in recent years, due to the need to manage a higher demand for health care and the introduction of new technologies, increased availability of treatments and provision of care across many settings [1]. These changes and demand on health care may make it difficult for health care organisations to manage care [1], for providers to deliver care, and may make it more burdensome for patients to receive and engage with their care [2–4]. One potential solution to these challenges is to consider and further develop models of care, such as care coordination [1]. Enhanced coordination is particularly beneficial for those with complex conditions such as chronic [2, 4] and rare conditions [3, 5–8]. Rare, ultra-rare and undiagnosed conditions are often complex, and affect multiple body systems and a person's mental and physical health [9, 10]. Rare conditions require care from multiple sectors, and health care professionals. For example, patients may be seen by primary care, secondary care, tertiary and quaternary care providers. Whilst individually rare conditions only affect a few individuals (each condition affects up to five in 10,000 people) [6, 9], collectively the 6000–8000 rare and ultra-rare conditions together with undiagnosed conditions affect a significant proportion of individuals worldwide [11].

Previous research has indicated that a lack of coordination has many negative impacts on patients and carers living with rare conditions, including on their physical health, finances, psychological wellbeing, and social aspects of their lives [12]. A recent scoping review of 154 reviews of common and rare chronic conditions, together with focus groups with patients, carers and health care professionals, has defined care coordination for rare conditions [13]. Findings indicated that coordination for rare conditions has many components and that there are many different options for how care can be coordinated [13]. Coordination was defined as everyone involved in a person's care (including the patient and/or family members) working together across multiple aspects of care to avoid duplication and achieve shared outcomes. Coordination would need to be lifelong and involve all parts of the health and social care system (including different services, settings, multiple conditions, and transition between services). It has been argued that coordination should be family-centred, evidence-based and ensure equal access for all [13]. The review highlighted many different components of coordination, including components that need to be coordinated (e.g., assessment), components that inform how to coordinate care (e.g.,

someone to take responsibility), components that have multiple roles (e.g., planning) and components that contextualise coordination (e.g., evidence-based practice) [13].

In order to better understand and potentially improve care coordination, it is necessary to identify and describe the different ways in which care can be coordinated for rare conditions. One way to facilitate the organisation of care coordination is to develop a taxonomy of care coordination for rare conditions. Taxonomies are systems used to organise complex concepts into common conceptual domains and dimensions based on similarities [14, 15]. Developing a taxonomy of care coordination for rare conditions would help to describe different ways of coordinating care. This in turn can facilitate the development and evaluation of pre-existing and new models of care coordination.

Taxonomies aim to provide clear definitions [15], and have been previously used to organise complex health care concepts including taxonomies of integrated care [16], health care [17], behaviour change [18], and the burden of treatment for patients with chronic conditions [19]. For example, the burden of treatment taxonomy included tasks that the health care system imposes on patients, factors worsening the burden of treatment and consequences of burden from the patients' point of view [19].

To the authors' knowledge, no previous studies have attempted to develop a taxonomy of coordination of care specifically for rare or chronic conditions. This study aimed to develop and refine a taxonomy of care coordination for people living with rare conditions, based on learning from the UK healthcare context and the National Health Service. Whilst there are many different rare conditions and the care needs for each of these may differ slightly, it is necessary to develop a taxonomy that can be used to outline the different options and then these options can be adapted and applied to suit different contexts and different rare, ultra-rare and undiagnosed conditions. We present our findings on what types of coordination may be appropriate in different situations and the development of hypothetical models of care coordination separately [20].

Methods

This study is part of a larger mixed-methods research project which aims to explore coordination of care for people living with rare conditions [21]. This study builds on previous aspects of this study [12, 13].

A summary of the methods used in this study are provided below (see Table 1 for a detailed description).

**Table 1** Detailed description of the methods used in this study

	Development of taxonomy (interviews and focus groups)	Refinement of taxonomy (workshops)
Design	- Previously both quantitative [22, 23] and qualitative [15, 19, 24, 25] approaches have been used to develop taxonomies. - Our study used qualitative methods (interviews, focus groups and workshops) and was conducted in a two-stage process (see Fig. 1). First, interviews and focus groups were conducted to develop an initial taxonomy. Workshops were then conducted to refine the proposed taxonomy. - Qualitative methods were used as they allowed for more in-depth understanding of coordination which is a multifaceted concept [24, 25]. Additionally, qualitative methods enabled us to involve stakeholders with the most experience of care coordination, which is particularly important in health care service research [26]. By understanding patients', carers' and health care professionals' views on the organisation of coordination of care for rare conditions it may be possible to improve health care services, and optimise the patient experience.	
Setting	- UK based study - Coordination across the NHS, social care and third sector (with a primary focus on health care)	
Sample	*Eligibility criteria* To participate in our study, participants needed to be: - 18 or over (Children were not included due to ethical issues recruiting participants under 18) - Patients with rare, ultra-rare or undiagnosed conditions, carers/parents of children or adults with a rare, ultra-rare or undiagnosed condition, health care professionals, charity representatives or commissioners *Recruitment methods* Participants were recruited using a range of methods, including: - Email invitation - Adverts on social media - Voluntary sector study advertisement - Adverts through our partnership with four NHS sites *Sampling criteria* To ensure that different models of coordinated care and a wide range of experience and expertise were captured, we purposively sampled using the following characteristics - For health care professionals/commissioners/charity representatives—area of the UK, job role, experience with different types of care coordination - For patients/carers—area of UK, condition, role, age, experience with different types of care coordination	
Measures	- To develop the taxonomy, we developed two topic guides: (1) interview topic guide and (2) focus group topic guide (Additional file 1: Appendix S1). - Questions focused on a range of topics including stakeholders' experiences of coordinated care, implications of coordinated care, preferences for aspects of care coordination (ways of coordinating care, format, access, frequency, location, information sharing, transition), benefits and challenges and factors that help or hinder coordination. - Feedback on the topic guide was sought from the CONCORD public and patient involvement advisory group prior to data collection.	- We developed one topic guide for the workshops (Additional file 2: Appendix S2). - The topic guide was based around the six categories identified in the taxonomy and included prompts on whether we had missed anything, whether findings seemed appropriate based on participants' experiences, appropriateness of options in light of the COVID-19 pandemic and recommendations to improve each of the six categories.
Procedure—recruitment and ethics	- Potential participants contacted the researcher (HW) via email or telephone and were given participant information sheets. - Potential participants were asked to provide responses to eligibility questions when registering their interest. - For professionals, these included: their occupation, speciality and geographical region. - For patients or carers, these included whether they receive coordinated care (specialist service and who coordinates), whether they have a diagnosis, are a patient/carer, their age range, ethnicity and geographical region. - Selected individuals were asked to complete two written consent forms prior to taking part in the interviews, focus groups or workshops. Participants who took part virtually or via telephone were asked to return written consent forms in advance. - Participants were informed that their data would be kept confidential, fully anonymised and that they could withdraw at any time without reason. Focus group participants were informed that any data collected up until the point of withdrawal would be kept due to difficulties removing individual participants from focus group data. We took steps to ensure that quotes from the participant who withdrew from the study were not included in publications.	

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Table 1 (continued)

	Development of taxonomy (interviews and focus groups)	Refinement of taxonomy (workshops)
Procedure—data collection	- One researcher (HW) conducted the interviews. - Interviews took place by phone (n = 27) or face-to-face (n = 3), depending on participants' preferences. - The interviews lasted approximately one hour (range 44–74 min). - Two researchers (HW and AS) conducted the four focus groups (one researcher facilitated, and one researcher took notes) [27]. A third researcher observed one of the focus groups (EH). Two focus groups were face-to-face in two cities in the UK, and two were conducted using Skype for Business. Focus groups were up to three hours in length (including a break) (range 149–154 min). - Interviews and focus groups were digitally recorded using an encrypted dictaphone (with consent from participants) and professionally transcribed. - Transcripts were checked for accuracy and fully anonymised (including names, places and due to their rareness—the names of specific conditions). - Data were stored in the university's secure data environment and coded using NVivo 12 [28].	- Workshop participants were sent a 15-min video prior to the workshop which outlined the findings of the taxonomy. - Participants were split into three breakout groups. - Each breakout group had one facilitator (HW, EH, AlGR) and one note taker (JJ, SM, AH). - After the breakout groups, participants reconvened in the main group and received feedback from each group on their discussions. - Workshops were recorded using an encrypted Dictaphone. - Notes were checked for thoroughness and summarised prior to being sent to a graphic facilitator (New Possibilities) to create a graphical representation of the findings (Additional file 3).

Table 1 (continued)

	Development of taxonomy (interviews and focus groups)	Refinement of taxonomy (workshops)
Analysis	- Thematic analysis was used to analyse interview and focus group data. In line with recommendations for taxonomy development [15]. - Inductive coding was used to develop an initial coding frame [29]. Six interview transcripts were coded inductively by two researchers (HW/AS). A coding framework was developed and agreed. The framework included codes on aspects of care coordination(types, who is involved, mode, information sharing, where, frequency, transition between services, methods of access) and qualifier codes (preferences, benefits and challenges, barriers and facilitators, factors influencing coordination). - The framework was used to code all interview and focus group transcripts (HW). A second researcher (AS) coded six interviews and one focus group transcript (20% of the data). Coding was discussed and any discrepancies (e.g. on how codes were used/what codes meant/when to use codes) were agreed. - Findings were then grouped into themes and sub-themes using thematic analysis [30]. Given the large amount of data, this was done in two stages; (1) development of themes and sub-themes for the data on aspects of coordination (to develop initial taxonomy options), (2) development of themes and sub-themes for the data on qualifying codes (to develop models) (described in [20]). - Five themes were developed (ways of organising care, ways of organising teams, responsibilities for coordination, access to coordination and mode of coordination). - Themes and sub-themes were discussed by co-authors and used to develop a taxonomy. - Once themes and sub-themes had been developed six stages of taxonomy development [31] were followed: (1) *Identify the meta-characteristic* that will inform the choice of characteristics in the taxonomy (2) *Identify ending conditions* (requirements that the taxonomy needs to meet to be finalised) (3) *Choose approach* We used an empirical-conceptual approach. We based the taxonomy on our findings from interviews and focus groups and earlier CONCORD findings (4) *Identify a subset of objects to classify*, using findings from the interviews and focus groups (5) *Identify common characteristics* (similarities and differences will be identified to identify common characteristics and discriminatory characteristics for coordinated care) (6) *Group the characteristics* using a manual process [14] - We reviewed our findings in relation to the CONCORD scoping review [13] and preliminary survey findings [21] to ensure that there were no major options missing from our taxonomy. - Additionally, the wider CONCORD team and CONCORD public patient involvement advisory group reviewed the taxonomy findings and provided feedback on the taxonomy prior to the workshop.	- Workshop notes were coded and grouped into themes surrounding their experiences of the model of coordination, benefits and challenges of the model of coordination, factors influencing coordination, missing aspects and impact of COVID-19. - Feedback on aspects that were missing in the taxonomy were used to refine and finalise the taxonomy. - Findings highlighted key aspects to be clarified within the taxonomy, including: the need to emphasise that care is not just medical (also includes social and educational aspects), and that care is lifelong. - Findings also highlighted the need to separate out collaborations that include patients/carers from collaborations between professionals. The need for third sector involvement in collaboration where appropriate; the need to emphasise the role that charities and patients/carers play in care coordination; a hybrid model of frequency and the need to clarify aspects of the mode domain. - The taxonomy was amended in line with this feedback.

Fig. 1 An overview of the two stages involved in this research
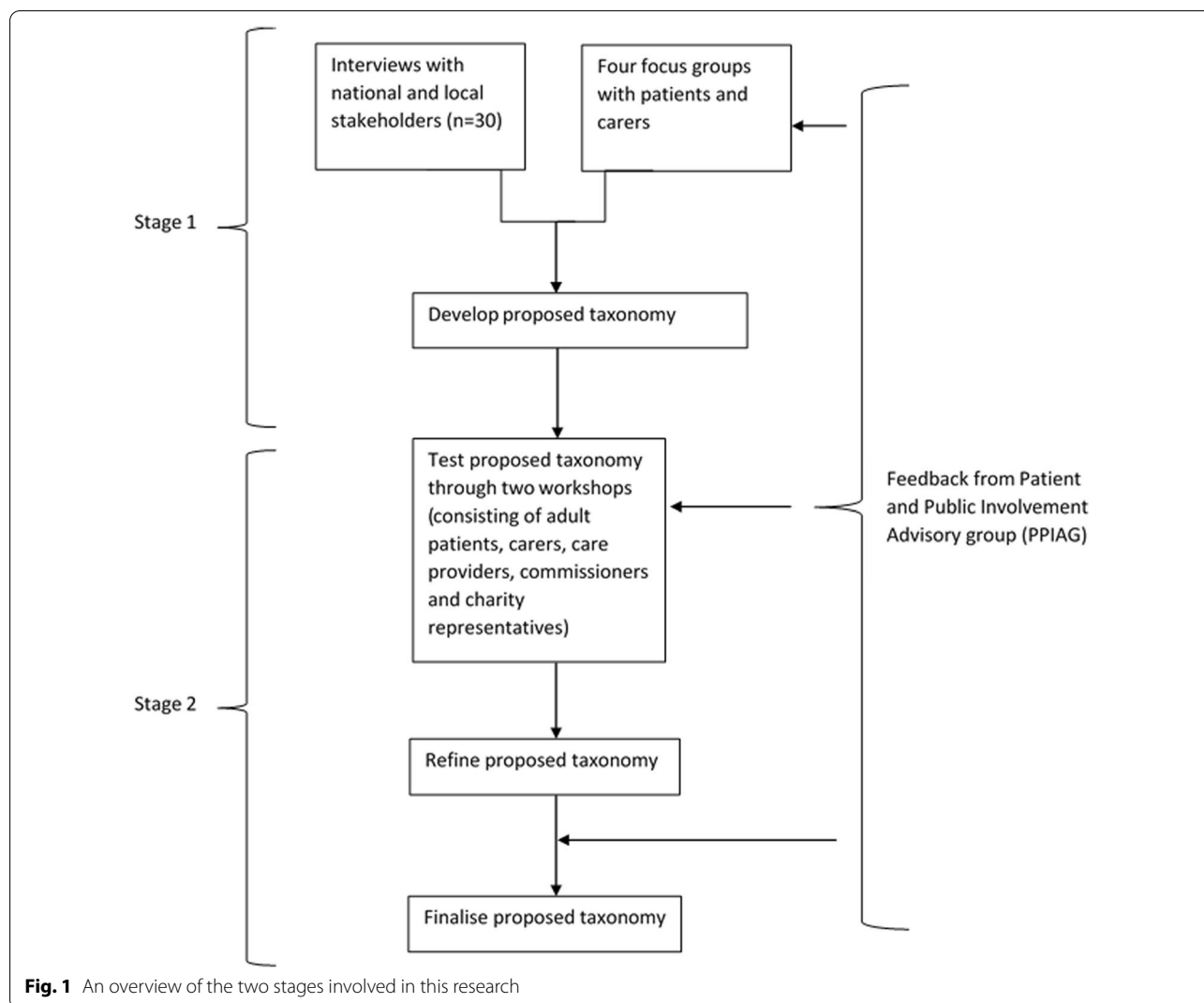
Design

A two-stage study using qualitative methods was conducted (see Fig. 1). In stage one, interviews and focus groups were conducted to develop a taxonomy. In stage two, workshops were conducted to refine the taxonomy.

Setting

Our study explored care coordination in the UK, across different sectors; with a focus on health care and the National Health Service.

Sample

Participants were recruited purposively using a range of methods including advertisements throughout the voluntary sector and advertisements through our partnership with four NHS sites. We recruited a range of individuals with experience of rare conditions,

including patients, parents/carers, and professionals (health care professionals, charity representatives and commissioners). Participants took part in interviews (n = 30 professionals), focus groups (four groups of 6–8 patients/carers) [27] and workshops (one patient/carer, and one professional; approximately 15 participants in each workshop). For patients and carers, we selected participants based on the area of the UK they lived in, their condition, role, age, and experience with different types of care coordination. For healthcare professionals, charity representatives and commissioners, we selected participants based on the area of the UK they worked in, their job role (to reflect a range of different job roles) and experience of different types of coordination. Eligibility criteria, recruitment methods and sampling criteria are provided in Table 1.

Procedure

Study adverts included details of the researcher's contact details and asked individuals to get in touch via email or telephone. Interested individuals contacted the researcher and were provided with an information sheet. Participants were asked to sign consent forms in advance of the interviews, focus groups and workshops. Participants were informed that their data would be kept confidential, fully anonymised and that they could withdraw without reason.

To develop the taxonomy, one researcher (HW) conducted interviews with health care professionals, commissioners and charity representatives. Two researchers (HW/AS), with a third observer (EH) conducted four focus groups with patients and carers (two face-to-face and two virtual). Interviews and focus groups were recorded with consent, professionally transcribed, checked for accuracy and fully anonymised. To develop the taxonomy and identify different ways of coordinating care, we used an interview topic guide and a focus group topic guide (Additional file 1: Appendix S1). See Table 1 for further details.

To refine the taxonomy, we held workshops with patients, carers, health care professionals, charity representatives and commissioners. We developed a workshop topic guide (Additional file 2: Appendix S2) to determine if the findings were appropriate and comprehensive. Workshop participants were sent a video outlining the findings prior to the workshops. Participants were split into three breakout groups and asked to discuss the findings for each of the six taxonomy domains. Workshops were recorded and notes were taken. Notes from the workshop were also used to produce a graphical representation of findings.

For a more detailed description of our procedure, please see Table 1.

Analysis

Thematic analysis was used to analyse interview and focus group data. In line with recommendations for taxonomy development [15]. Inductive coding was used to develop the coding framework [29]. Two researchers (HW/AS) initially coded six interview transcripts to develop and agree a coding frame. The coding framework was then used to code all interview and focus group transcripts by one researcher (HW). A second researcher (AS) also coded 20% of the data. Findings were discussed and agreed. Findings were then grouped into themes and sub-themes. Given the large amount of data, this was done in two stages: (1) development of themes and sub-themes for the data on aspects of coordination (to develop initial taxonomy options), (2) development of themes and sub-themes for findings relating to which models of care coordination work in different situations (described in [20]). Themes were discussed by co-authors and used to develop a taxonomy. Findings were also used to explore which models of care coordination may be appropriate in different situations and to develop hypothetical models of care coordination (these findings are described elsewhere in [20]).

Once themes and sub-themes had been developed, we followed six stages of taxonomy development to develop the proposed taxonomy [32]: (1) Identify the meta-characteristic (focus of the taxonomy), (2) Identify ending conditions, (3) Choose approach, (4) Identify a subset of objects to classify, (5) Identify common characteristics and (6) Group the characteristics [14]. Table 2 outlines how we applied these six steps.

Table 2 Description of how we applied Nickerson et al.'s [31] taxonomy development criteria

Step	Our process
1. Identify meta-characteristic	Meta characteristic = different ways in which care can be coordinated for rare conditions
2. Identify ending conditions	Our ending conditions: 1. Not merging or splitting any objects in the last iteration 2. Having at least one object (type of coordinated care) under every characteristic of every dimension 3. Not adding any new dimensions or characteristics in last iteration 4. Uniqueness of dimensions, characteristics and cells
3. Decide on approach	We used an empirical-conceptual approach. We based the taxonomy on our findings from interviews and focus groups and earlier CONCORD findings
4. Use a subset of objects to classify	We used themes and sub-themes from the interviews and focus groups as objects to classify. The sub-themes outline types of coordination that can be used as objects (e.g., nationally commissioned services and condition-specific clinics). List of 'objects' (example ways of coordinating care) were identified from themes and sub-themes
5. Identify common characteristic	Similarities and differences were identified to identify common characteristics and discriminatory characteristics. These were identified through the summaries of themes and sub-themes
6. Group characteristics using a manual or graphical process	We used a manual process to group characteristics into domains to form the first draft of the taxonomy

To refine the taxonomy, we coded workshop notes and grouped them into themes (see Additional file 3: Appendix S3 for a visual representation of workshop findings). Findings were used to amend the taxonomy. See Table 1 for further details.

Results

Participant characteristics

This study included 77 different participants (two participants took part in both a workshop and interview/focus group). Participants included patients, carers, health care professionals, commissioners and charity representatives. Overall, data from 52 participants (30 individual interviews, 22 focus group participants) informed the development of the taxonomy. Data from 27 workshop participants informed the refinement of the taxonomy. Demographic characteristics are shown in Table 3.

Whilst it would not have been possible to represent all 6000–8000 conditions within our study, the patients, carers, healthcare professionals, charity representatives and commissioners that took part in our study were selected to represent a range of different rare conditions (including different characteristics, presentations, and models of care coordination). Additionally, professionals, charity representatives and commissioners were able to draw upon experiences of working across different types of rare conditions. Examples of groups of rare conditions included in this study included rare skin conditions, rare chromosome conditions and rare autoinflammatory conditions. We sampled a range of different rare, ultra-rare and undiagnosed conditions to ensure that our sample was as representative of care coordination for rare conditions as possible. We included a range of different professionals with different job roles in our study, including health and social care professionals (e.g. consultants from different disciplines, allied health professionals, genetic counsellors, pharmacists, psychiatrists, GPs), charity representatives (with various roles) and national and local commissioners (see Table 3 for details).

Taxonomy of care coordination for rare conditions

Our final taxonomy of care coordination consists of six domains: (1) ways of organising care; (2) ways of organising those involved in a patient's care; (3) responsibility for coordination; (4) how often appointments and coordination take place; (5) access, and (6) mode (see Table 4). Each domain outlines different ways of coordinating care (labelled 'sub-domains'). Within each way of coordinating care there are different options (labelled 'options').

Within the next section we will present each of the taxonomy domains and their sub-domains in turn. Example quotes for each of the six domains are shown in Table 5.

Domain 1. Ways of organising care

Our findings highlighted different ways of organising care. Options ranged from local care provision where all care is delivered locally, through to care being delivered in a single national centre that serves all patients in the country with a particular rare condition. Figure 2 provides a summary of the different ways of organising care.

Nationally centralised

For nationally centralised services, we identified different examples of nationally centralised care where care is delivered or coordinated centrally. Central delivery of care can either take place in one nationally commissioned centre/service (such as rare disease centres or condition-specific centres) or a network of multiple services or centres.

Hybrid

We also identified some 'hybrid' options, which combine both national or regional specialist and local care. Hybrid options include hub and spoke networks, and outreach. There are different types of hub and spoke models. For example, in one type of hub and spoke model of care, the national centre or centres (hub) coordinate care but the actual care delivery happens at a local hospital or GP (spokes). In other types of hub and spoke models, the national centre (hub) provides some care delivery, but other aspects of care are delivered at local hospitals or GPs (spokes). There are also different types of outreach models. Examples of outreach models include outreach clinics (e.g. local outreach clinic, specialists travelling to provide joint clinics with local team, specialists providing care locally) and outreach relating to care coordination (e.g. outreach model of clinical case management in mental health practice, coordinator doing outreach work with local providers and GP and coordinator travelling to provide care locally). Outreach models relating to education included specialist teams providing support for local teams (e.g., education to raise awareness, providing guidance and supervision, email hotline, training, opportunities for local providers to shadow clinics, and formalised agreements that specialists will answer GP queries).

Local

Findings highlighted the importance of specialists being involved in care for rare conditions. However, findings also indicated that routine care and non-standardised or tailored care should be delivered locally, and that regular contact with local professionals would be useful. On the other hand, some focus group participants reported wanting all of their care to be delivered locally, or regionally. For some, there was a lack of local care provided.

Table 3 Demographic characteristics of participants

	Development of taxonomy (n = 52)		Refinement of taxonomy (n = 27)		Total
	Interviews	Focus groups	Patient and carer workshop	Professional workshop	
Number of participants	30	22[a]	12	15	79 (77 different people[b])
Type of participant					
Patients	N/A	16	5	N/A	21
Parents/carers of children aged < 18 years	N/A	5	4	N/A	9
Parents/carers (e.g. spouses) of adults aged ≥ 18 years	N/A	1	3	N/A	4
Health care professionals[c,h]	15	N/A	N/A	2	17
Health care professionals employed by charity	2	N/A	N/A	2	4
Charity representatives[d,h]	5	N/A	N/A	8	13
Commissioners	3	N/A	N/A	3	6
Multiple professional roles[e]	5	N/A	N/A	N/A	5
Age (years)					
18–25	N/A	2	0	N/A	2
26–59	N/A	16	10	N/A	26
≥ 60	N/A	4	2	N/A	6
Diagnosis[i]					
Rare/ultra-rare condition(s)	N/A	22	12	N/A	34
Attend specialised service[f]					
Yes	N/A	14	6	N/A	20
No	N/A	7	4	N/A	11
Not sure	N/A	1	2	N/A	3
Locations represented					
National role (UK)	2	0	0	8	10
National role (England and Wales)	1	0	0	1	2
National role (England)	5	0	0	3	8
Scotland	1	0	1	0	2
Wales	1	1	0	0	2
East of England	1	2	1	1 [g]	5
London	4	7	0	0	11
Yorkshire and the Humber	1	2	0	0	3
North East of England	1	2	0	0	3
North of England	1	0	0	0	1
North West of England	2	3	1	0	6
South East of England	1	2	3	0	6
South West of England	4	0	4	1	9
West Midlands	5	2	1	1	9
East Midlands	0	1	1	1 [g]	3
Ethnicity					
White	N/A	19	12	N/A	31
Other	N/A	2	0	N/A	2
Not specified	N/A	1	0	N/A	1
Who coordinates care?					
Patient/carer	N/A	17	10	N/A	27
GP	N/A	1	0	N/A	1
Member of health care team	N/A	1	0	N/A	1
GP and patient/carer	N/A	2	1	N/A	3

Table 3 (continued)

	Development of taxonomy (n = 52)		Refinement of taxonomy (n = 27)		Total
	Interviews	**Focus groups**	**Patient and carer workshop**	**Professional workshop**	
Other	N/A	1	0	N/A	1
Don't know	N/A	0	1	N/A	1

N/A, not applicable as patients/carers and health care professionals were asked different eligibility questions

[a] Initially had 23 participants but 1 withdrew their data post focus group

.[b] Two of the interview participants also took part in the workshops

[c] A range of health care professionals were included within our sample. Job roles included: consultant (various specialities), specialist nurse, GP, allied health professionals (speech and language therapist, physiotherapist, occupational therapist), genetic counsellor, pharmacist, coordinator, psychiatrist

[d] Charity representatives were from a range of charities which represented patients with rare conditions

[e] Some of the participants had multiple roles within the professional category, e.g. being a health care professional and a commissioner, or being a health care professional and a charity representative

[f] We asked participants if they attended a specialist service or not. Responses may include seeing specialists in their condition in addition to specialist services

[g] Role covers both locations

[h] A few health care professionals/charity representatives also had personal experience of rare conditions as patients/carers

[i] Although people with an undiagnosed condition were eligible to take part, none participated

Domain 2. Ways of organising those involved in a patient's care

Our findings highlighted different ways of organising those involved in a patient's care (including professionals involved in a patient's care, the patient and their family). This includes two types of collaboration: collaborative working between professionals, and collaborative working between professionals, patients and carers. Options ranged from collaboration between many or all of the individuals involved in a patient's care, to collaboration between some of the individuals involved in a patient's care, to a lack of collaborative working (see Table 4 for examples). Workshop findings highlighted that COVID-19 may have offered new opportunities for collaboration, such as the ability for local team members to dial into multidisciplinary team meetings. Figure 3 provides a summary of the different ways of organising teams.

Collaboration between many or all of those involved in a patient's care

Examples of collaboration between many or all of the individuals involved in a patient's care includes condition specific clinics (for example those organised and led by individual health care professionals and those delivered as rare disease or specialist clinics) and multidisciplinary meetings held between all professionals (and the patient/carer where appropriate). Different types of condition-specific clinics exist, ranging from: multidisciplinary team appointments including all professionals (and the patient/carer where appropriate); one stop shops where patients receive all care in one place at the same time;

multidisciplinary clinics that involve professionals seeing patients both together as a team but also separately; and carousel clinics whereby the health care professional moves around whilst the patient stays in the same room.

Collaboration between some of those involved in a patient's care

One example of collaboration between some of the professionals involved in a patient's care is joint clinics. Joint clinics consist of a couple of professionals working together to provide care. For example, joint clinics consisting of an adult and a child provider; joint clinics consisting of multiple consultants; and joint clinics consisting of specialist and local providers. Additionally, close working between different professionals may occur (e.g. paediatrician contacting adult provider when the patient is ready to transition to adult care).

Lack of collaborative working

In some cases, examples demonstrating a lack of collaborative working between those involved in a patient's care were identified, including a lack of multidisciplinary team clinic, lack of transition methods and lack of ownership.

Domain 3. Types of responsibilities and roles needed for care coordination

Our findings highlighted different types of responsibility and roles involved in coordinating care for rare conditions (administrative, formal and supportive roles) across health care, social care and voluntary sectors. Figure 4 provides a summary of the different types of responsibility and roles.

Table 4 Taxonomy of care coordination for rare conditions

Domain	Sub-domain	Options	Examples
1. Ways of organising care	Local	Local care delivery	All care delivered locally—in one place, or multiple places—including hospital and home visits, emergency care
		Local care coordination	All coordination delivered locally—e.g. coordination appointments local to the patient
	Hybrid (combination of specialist and local) (e.g. hub and spoke models)	Coordination nationally centralised but delivered locally	Specialist service coordinating care but care delivery is done locally (e.g. at local hospital or GP)
		Care nationally centralised but delivered locally	Care nationally centralised with outreach, specialist providers with routine care from local providers
		Types of outreach models	Outreach support for professionals, outreach clinics, outreach care coordination, outreach education
		Regionally centralised care	Regional network models, regionally delivered services
	Nationally centralised	Care delivered and coordinated centrally	Specialist centre, rare disease centre or service
		Care delivered centrally (in one nationally commissioned service or centre)	Nationally commissioned service or rare disease centres, adult and paediatric centres or condition specific centres
		Care delivered centrally in multiple services/centres or as part of a network	National network models to deliver care and coordination and share expertise, nationally commissioned services
2. Ways of organising those involved in a patient's care (including professionals, patient and/or carer)	Lack of collaborative working between professionals involved in a patient's care	Professionals not working together (health care, social care, third sector if appropriate, etc.)	Lack of multidisciplinary team (MDT) working, lack of collaborative working
	Collaboration between some of the professionals involved in a patient's care	Some professionals working together to provide care (health care, social care, third sector if appropriate, etc.)	Joint clinics with specialist and local providers or adult and paediatric providers
		Continuity of professionals	Same professionals throughout care, professionals attending appointments with patients
	Collaboration between many or all professionals involved in a patient's care	All professionals working together to provide care (health care, social care, third sector if appropriate, etc.)	Condition specific clinics—run by health care professionals, within specialist service, one stop shop, carousel clinic
		All professionals meeting together to discuss care (health care, social care, etc.)	MDT meeting, or health care professionals attending Education, Health and Care Plan meetings
	Lack of collaborative working between professionals and patients/carers	Professionals not working with patients	Lack of collaboration with patients (e.g. lack of involvement in MDT meetings)
	Collaboration between some professionals and patients/carers	Professionals working with patients to prepare them	Orientation visits/transition events/advice and support
		Patients meeting to discuss care	
	Collaboration between many or all professionals involved in a patient's care and the patient/carer	Professionals meeting together with patient/carer (health care, social care, third sector if appropriate, etc.)	Patient involvement in MDT meeting where appropriate

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Table 4 (continued)

Domain	Sub-domain	Options	Examples
3. Responsibilities	Administrative support	Administrator	A combination of an administrator and the patient and carer (e.g. working together to arrange appointments)
			An administrator/service PA or secretary (e.g. to produce letters and plans, take calls, organise clinics, act as the first point of contact for patients and update GPs).
			Rare disease charities (e.g. to provide administrative support, support with travel arrangements and answering queries)
			Automated support (e.g. a hospital appointment system)
		Point of contact for patients	Clinicians (e.g. consultants, nurses, community matrons, coordinators, geneticists, medical social workers, or disability nurses)
			Administrators (e.g. secretaries)
			Charity workers (e.g. charity patient support workers) and youth workers
		Point of contact for professionals (health care, social care, etc.)	Coordinator, specialist
	Formal roles/responsibilities	Administrative coordinator	Clinic coordinator—could be range of roles, including patient/carer, non-medical professional, charity employed support worker, nurse or allied health professional equivalent
		Care coordinator	Someone with system and condition knowledge such as a nurse or allied health professional equivalent or hospice/community nurse / social care professional / non-medical professional / charity employed support worker / transition coordinator / doctor equivalent role
		Clinical coordinator	Someone with sufficient clinical expertise to coordinate complexity—doctor equivalent role, GP
		Clinical lead	Someone with oversight over care such as a nurse, doctor equivalent role, GP
		GP	Coordination, and implementing care plans from specialist
		Charities / patient support networks (in some situations)	Direct roles in coordination (e.g. clinic coordinators/coordinating care), supporting coordination and advocating on patients' behalf
	Supportive roles	Charities / patient support networks	Direct roles in coordination (e.g. clinic coordinators/coordinating care), supporting coordination and advocating on patients' behalf
		Patients and carers	Direct role as coordinators, providing education to professionals, part of the MDT and information provision
		Peers	Providing support for coordination
	No responsibility	No point of contact / coordinator / clinical lead / GP / no hospital ownership	

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Table 4 (continued)

Domain	Sub-domain	Options	Examples
4. How often care appointments and coordination appointments take place	Regular	Care appointments	*Ranging from multiple times per week—weekly—every 3 months—annually*
		Coordination appointments	*Ranging from more than once a month—monthly—every 2 months—every 6 months—annually*
		Meetings	*Ranging from before every clinic—weekly—twice a month—monthly—every 3 or 4 months –every 6 months- annually*
	On demand—when needed	Care appointments	*On demand care appointments, coordination or specialist centre appointments when needed*
	Hybrid (combination of regular and on demand)	Regular appointments (as above) with on demand in between as and when needed	*Regular appointments but with on demand appointments (care appointments, coordination appointments or specialist centre appointments) as and when needed*
5. Access to records	Full access	Health care professionals	*Health care professionals having full access to records*
		Patients and/or carers	*Patients and/or carers having full access to records*
	Filtered access (information filtered to necessary information that is needed by the relevant individuals)	Health care professionals	*Health care professionals having access to the relevant necessary information that is needed*
		Patients	*Patients and/or carers having access to the relevant necessary information that is needed*
		Third sector (where deemed necessary)	*Charity organisations having access to relevant necessary information if needed (e.g., when involved in care delivery/ coordination)*
6. Mode[a] of contact	Digital	Information sharing	*Digital records, digital letters, digital databases and registries, digital portals, mobile applications for patients and digital patient information*
		Coordinated care delivery	*Video appointments with professionals, virtual MDT clinics, digital ways of tracking symptoms e.g. electronic wearable devices, virtual tours of wards, apps to record test results, diagnostic technology, virtual centres*
		Coordination	*Video appointments with coordinator, coordination in the cloud, virtual review (as lowest level of coordination)*
		Communication (between professionals)	*Virtual panels to discuss cases with experts, email hotlines, virtual MDT meetings and clinics, email contact*
		Communication (between professionals, patients and carers)	*Email contact*
	Face-to-face	Coordinated care delivery	*Initial meetings, key treatment phases such as diagnosis and stabilisation, physical exams, clinic appointments, home appointments*
		Coordination	*Face-to-face meetings between patients and coordinator*
		Communication (between professionals)	*Face-to-face team meetings*
		Information sharing	*Via coordinator and meetings*

Table 4 (continued)

Domain	Sub-domain	Options	Examples
	Telephone	Coordinated care delivery	Telephone clinics and consultations, conference calls, appointments such as GP appointments, telephone calls when needed, discharge calls and follow-up appointments
		Coordination	Telephone calls with coordinators, initial introductions, coordination of care via phone, NHS 111 style phone service to coordinate care for rare conditions, WhatsApp contact with coordinator
		Communication (between professionals)	Phone calls with other professionals, contacting specialists, professional conference calls, discussing treatment plans, asking local teams to implement care plans
		Communication (between professionals and patients/carers)	Telephone advice services or direct line to team, regular check-ups, phoning departments, WhatsApp contact, phone calls between patient and professionals, messaging peers
	Written	Information sharing—care documentation	Written records such as condition specific passports and alert cards Written letters such as clinic letters, discharge letters and summary letters Care plans for patients such as agreed care plans, shared care protocols, Education Health Care Plans, transition plans Reports such as written reports and handover packs and transition reports and booklets and Summary of records
		Information sharing—service planning	Plans to specify hospital and health care professional roles and responsibilities Standard operating procedures to record MDT working
		Information sharing—guidelines and care pathways	Service specifications Quality assurance standards Governance frameworks National guidelines such as NICE, charity produced, or specialist service produced International best practice Lack of evidence-based pathways For coordinators
		Information sharing—training policies and frameworks	For coordinators, supervisors
	Lack of (communication mode)	Information sharing	Lack of letters, care plans
		Communication	Between professionals or professionals and patients

Examples given in this taxonomy refer to those identified throughout interviews, focus groups (and then validated within the workshops). Some of these examples may be in practice currently and some of which are ideas for new ways of coordinating care

'Care' refers to all aspects of care, including both health and social care. Care also refers to lifelong care (including transition from paediatric to adult services)

Findings relating to where care is coordinated/delivered have been combined with way care is organised (domain 1)'—as there is lots of overlap

a Modes can be combined. We identified many examples of combined modes in practice (e.g. face-to-face and digital, face-to-face and phone, digital and phone or face-to-face, phone and digital)

Table 5 Example quotes (from interviews and focus groups) for each of the six domains

Domain	Sub-domain	Example quote
Ways of organising care	National	"Yeah, we've been running our multi-specialty clinics for about 18 months now in our new Rare Disease Centre" (interviewee, health care professional)
	Hybrid	"So, [Place 3] is our lead paediatric centre, so they see all the local [Place 3] patients, and they are our hub, we are a spoke, so we look after the patients locally in [Place 2]. But [Place 3] very much do like the guidelines that we follow and everything like that, and they are available to contact […] and like I said once a year they will see every patient in our clinic" (interviewee, charity representative and health care professional)
	Local	"I live in deepest darkest, it's rural [Region 1], nearly as far away from the central hospitals of [Place 3] and [Place 2] as you can get. So I want all my care in the community and that of my son, I want everything down here, because you know, there's no public transport, there's no, I mean, literally there are no buses where we live, anywhere. To get anywhere, yeah, there's just nothing. And so we need something that is definitely in the community, and also communities can be very different" (interviewee, patient group representative)
Ways of organising those involved in a patient's care (including professionals and patient and/or carer)	Collaboration between many or all of those involved	"The [rare condition x] clinic does try to address some of those deficiencies by providing a platform for coordinated care. […] they can come to the clinic here and see six different specialties simultaneously, and those different specialties can then try and formulate a care plan which incorporates aspects of each specialty's contribution" (interviewee, health care professional)
	Collaboration between some of those involved	"But what we try to do is to ensure that there is a joint transition clinic between the paediatrician and the receiving adult clinician and a visit to the hospital, which is usually supported […] by one of the workers from the children's unit" (interviewee, commissioner)
	Lack of collaborative working	"My experience currently of coordinated care is that there is none. It sounds like a complete and utter fantasy to me" (focus group participant, parent/carer)
Responsibilities	Administrative support	"We've got an admin person and she's quite instrumental at helping us set those up as well […] so that's a useful, really useful resource that we have" (interviewee, health care professional)
		"Yeah, we have a—when a patient is new to the service they'll get given quite a lot of contacts, including our health email" (interviewee, health care professional)
	Formal responsibilities	"there could be a stratified level of lead with a, sort of, triangle, an upturned triangle with a base at the bottom, the pinnacle at the top, and then, actually, the other way around, that the digital is at the bottom along with the smallest amount of care, and then, you know, you might have a patient requiring, you know, a quarterly or even a monthly telephone call with the coordinator or the community nurse, or whatever. […] Certainly, you start with digital and then you would have a monthly phone call or a quarterly phone call depending on what the anticipated need of that patient is, and then it could be escalated up as required" (interviewee, commissioner)

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Table 5 (continued)

Domain	Sub-domain	Example quote
		"I guess it's fairly, sort of, just everyone, sort of, chipping in, but I guess, obviously, the consultant's there and, ultimately, they will try and… You know, if we're struggling with it, then they might, sort of, take more control of that conversation and be, like- or suggest, "Why don't you do it like this?" but, generally, it's, kind of, us just, sort of, negotiating between ourselves" (interviewee, health care professional)
		"I think that a GP is the closest thing I have to a care coordinator […] feel like they might be best equipped to sort of coordinate care if they had more time and training to do it or even budget to do it" (focus group participant, patient)
	Supportive roles	"but they [patient support groups] are very good at picking up the pieces, supporting patients and providing information that the health care professionals don't provide, so they're key I think" (interviewee, health care professional)
		"I'm pretty much [Name 1]'s care co-ordinator. She sees about 15 to 16 different specialists" (focus group participant, parent/carer)
How often care appointments and coordination take place	Regular	"so there could be kind of like different levels of how often you need to see people, but I think definitely for us it would be that it would be ongoing at the minute" (focus group participant, parent/carer)
	On demand	"I find sometimes if you have yearly or six-monthly appointments time and time again, they can be a bit fruitless" (focus group participant, patient)
Access to records	Full access	"Well, that gets us back to the electronic patient record, doesn't it? you know, ideally, I think there should be an electronic patient record that is accessible to everyone involved in someone's care. Unless that is available, communication always ends up as a weak link, doesn't it?" (interviewee, health care professional) "I just want it to be shared with me, and it can't, and they never let you see everything" (focus group participant, patient)
	Restricted access	"Yeah, so in essence, the way…what I've just really said, I think the information needs to be available to all who need to have it, obviously with appropriate restrictions" (interviewee, health care professional) "I would like something like that on my health records of who wants to look at it, with a little bit of why, then yes, I'll just tick yes, but also, I'd like a list of who has accessed it. […] Because I want to know who's reading my, you know, someone did say at one time, "Oh, the psychiatric team are looking at your notes," I haven't given them permission to do that. […] You know, why are they looking at my notes and for what reason?" (focus group participant, patient)
Mode of contact	Information sharing	"Well it is having it, so basically so there is communication from one place to thenext. […] if everything's joined up beautifully electronically, that'll be there anyway almost" (interviewee, health care professional)
		"it's really helpful that there's a sort of overarching operating policy or operating manual for any service" (interviewee, commissioner)

Table 5 (continued)

Domain	Sub-domain	Example quote
	Care and coordination appointments	"there needs at least to be a connection with a multidisciplinary physical structure […]. And otherwise the coordination of care could also be digital, as we said beforehand. You know, it could be on the cloud" (interviewee, health care professional)
		"a new diagnostic result. I think this requires face-to-face contact with, you know, an expert or a coordinating clinician. This is, you know, it's like giving someone a new name. So, I think it is very important that there's a face-to-face contact with a medical professional when this happens. Then I think there is a need for face-to-face contact when there's a new kind of clinical or medical complication, but that face-to-face contact need not necessarily be with the coordinating clinician; that could be with the relevant clinician" (interviewee, health care professional)

Administrative support

Administrative support for appointment organisation was highlighted as important. Additionally, having a point of contact for patients and having a point of contact for other professionals was felt to be important. Some patients and carers highlighted that they do not currently have a point of contact. Different options were highlighted in relation to who should provide administrative support (e.g. administrator and patient/carer, administrator, charities, automated support) and who should be the point of contact for patients (e.g. clinicians, administrators, charity workers).

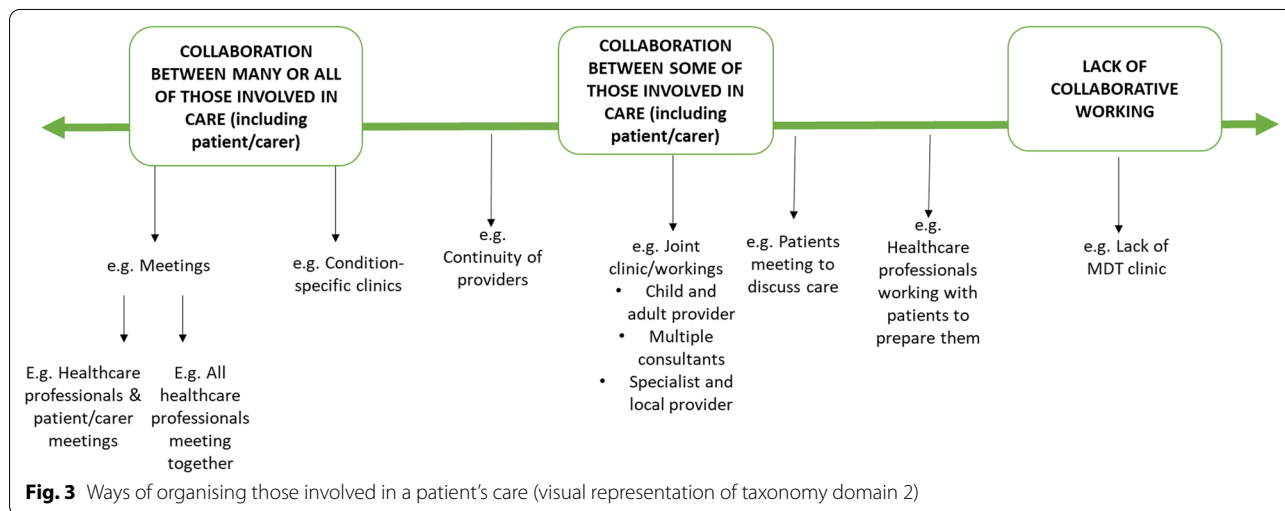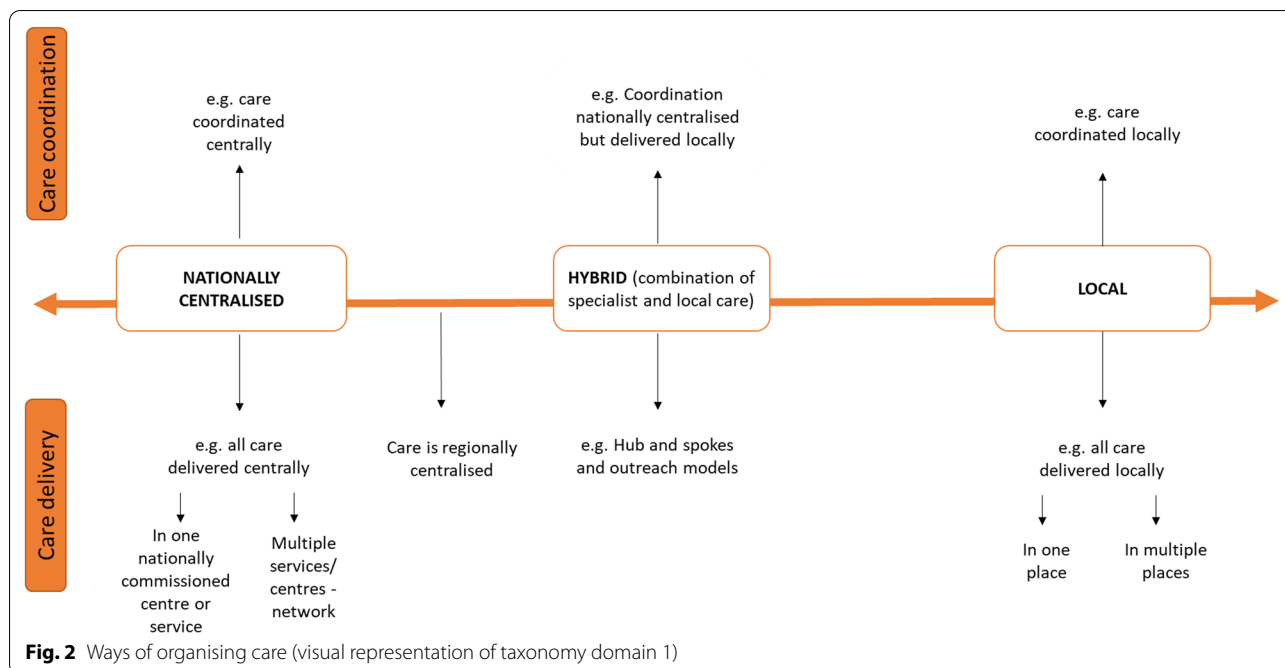### Formal coordination responsibilities

Formal coordination responsibilities across three roles were identified: (1) those conducted by a coordinator, (2) those conducted by a clinical lead in secondary/tertiary care, and (3) those conducted by a GP in primary care.

Our findings highlighted many roles of a care coordinator (e.g., liaising with health care professionals, coordinating the MDT and aspects of care across different sectors, trusted named person for the patient, ownership, quality assurance and planning). Many different types of coordinators were identified: (1) administrative coordinators, (2) care coordinators and (3) clinical coordinators. Administrative coordinators are individuals who arrange MDTs and clinics (e.g. patient/carers, non-medical professionals, charity employed social workers, nurse or allied health professionals). Care coordinators are individuals who have a formal/professional role for coordinating care in addition to system and condition knowledge (e.g. specialist nurses, allied health professionals, hospice/community nurses, social care professionals, non-medical professionals, charity employed social workers and transition coordinators). Clinical coordinators are individuals with sufficient clinical expertise to coordinate complex cases (e.g. doctors or GPs).

Our findings highlighted many roles of a clinical lead, including overseeing or managing care in a service, clinical case management, supervision of professionals, decision-making about extent to which different levels of coordinator are needed, and delegating and ensuring accountability of responsibilities. Clinical leads were identified from a range of roles (e.g. consultants, discipline-specific clinical leads, paediatricians (e.g., hospital/community), the patient's favourite doctor, a specialist nurse, or geneticists). Some patients and carers reported that they did not have a clinical lead responsible for their care.

Findings highlighted that GPs may have the potential to be involved in coordination in numerous ways, including as a coordinator (e.g. making appointments, named coordinator, developing care plans, identifying when patients need to see coordinator), clinical lead (e.g. a role between primary care and tertiary care to enable them to be responsible for care, or having a GP equivalent role for rare conditions), and implementing care plans provided by specialists (e.g. gatekeeper for specialist care referrals, providing local care and implementing care plans). However, findings indicate that many GPs do not take ownership and that some patients do not have a named GP. A lack of communication between GPs and specialists indicated a need for further collaboration between GPs and specialists (e.g., involvement in MDTs, working in hospital settings, receiving training by specialists, formalised contracts, point of contact).

Fig. 2 Ways of organising care (visual representation of taxonomy domain 1)

Fig. 3 Ways of organising those involved in a patient's care (visual representation of taxonomy domain 2)

Supportive roles

Supportive roles were also identified, including those conducted by charities/patient organisations and carers and those conducted by patients/carers (see Table 4).

Findings indicated that charities/patient organisations play many roles in coordination including supporting coordination by providing support to patients/carers (e.g. providing information, holding support groups, providing helplines) and professionals (e.g. training of professionals and guiding coordinators). Charities also have direct roles in coordination (e.g. funding coordinator posts and clinical networks, being clinic coordinators,

coordinating care, providing resources to help coordinate care, and managing registries). Whilst charities provide funding currently there were views that charities should not be filling gaps for health care services and that charities may not have the capacity to be the main coordinator. Additionally, charities play a role in service quality and improvement (e.g., care pathway development, pulling together evidence, identifying weaknesses in coordination, and setting up/developing specialist services).

Findings indicated that patients and carers currently have lots of direct involvement in coordinating care. Patients and carers act as coordinators of care (e.g.

Fig. 4 Types of responsibilities for coordination (visual representation of taxonomy domain 3)
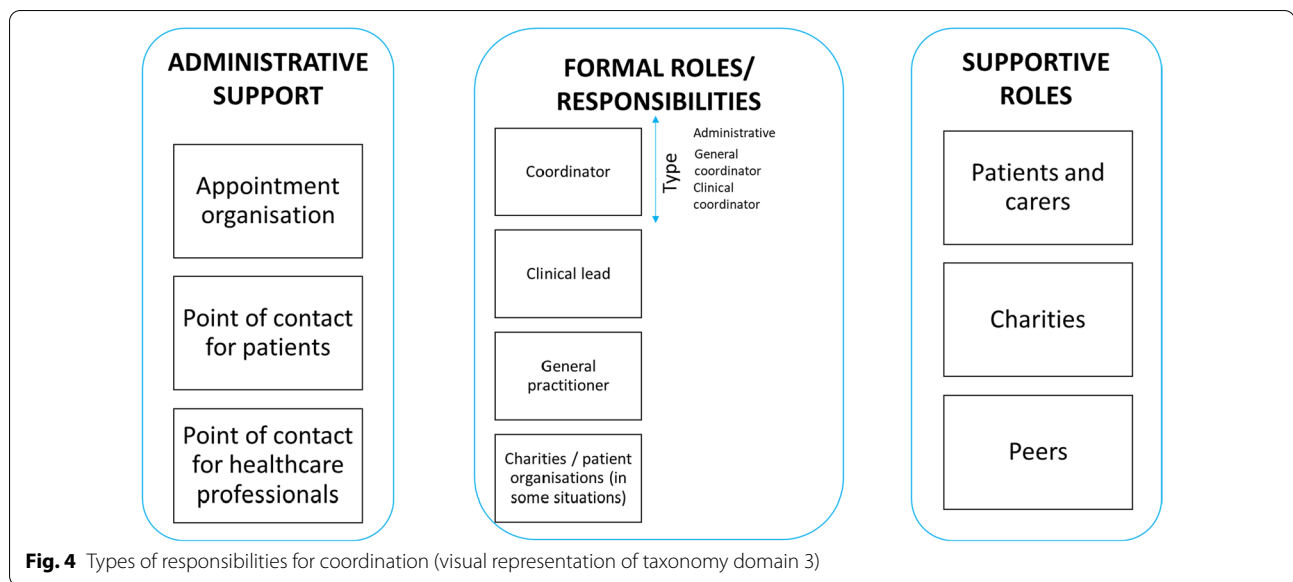
coordinating care across multiple hospitals, being their own advocates, taking more responsibility and chasing appointments) or collaborate with health care professionals to coordinate care (e.g. to arrange appointments, support from health care professionals for coordination as and when needed, and wanting care to be coordinated in partnership with themselves). Patients and carers also support transition to adult services, provide education to health care professionals, and monitor their own care.

Domain 4. How often care appointments and coordination take place

Our findings highlighted different time periods for care appointments and coordination activities. Options included regular appointments, on demand appointments and hybrid of regular and on demand appointments (see Table 3). Workshop findings highlighted that COVID-19 may have provided more opportunities for on demand appointments for those with stable conditions. See Fig. 5 for a summary of this domain.

Regular

Regular appointments were discussed in relation to: frequency of regular specialist centre appointments (ranging from six weeks post treatment to every 18 months), frequency of regular care appointments (ranging from multiple appointments in one week to every 6 months), frequency of contact (ranging from monthly check-ins to yearly check ins), frequency of outreach visits (e.g. annually), frequency of contact with coordinator (ranging from monthly to annually) and frequency of MDT meetings between health care professionals (ranging from weekly to twice a year). Participants indicated that

the regularity of appointments is and should be guided by condition-specific guidelines (where available).

On demand

Findings from some focus group participants and some interviewees indicated that on demand contact or care appointments (with specialists and coordinators) may be better than regular contact for some patients. One caveat to on demand appointments was that there needs to be quick access to expertise and care in emergencies.
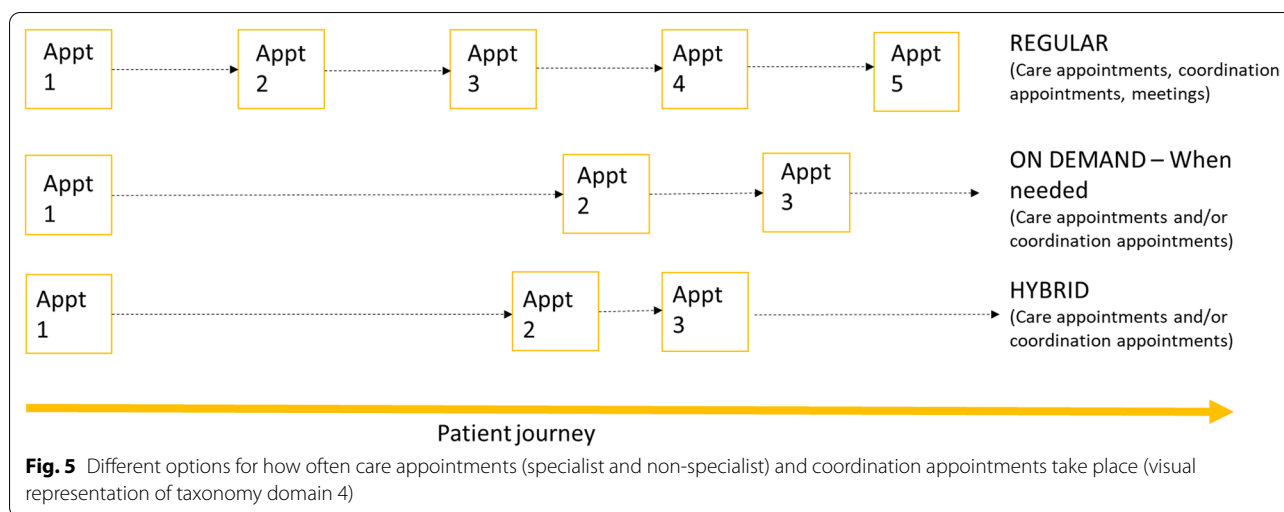
Hybrid

Workshop findings highlighted the need for a hybrid category that combines both regular care (at a minimum) with on demand support.

Domain 5. Access to records

Our findings highlighted different types of access. Options ranged from full or restricted access to records for patients and providers and access to support for patients and health care professionals (see Table 4).

For patients, findings highlighted the need for patients and carers to have access to their own information. This included: access to their records, and access to meetings concerning them.

For health care professionals, findings indicated the importance of access to information and records, given that patients are often seen in different places and by different professionals. The extent to which health care professionals should access information and records was contested; with some patients and carers thinking that any health care professional involved in care should be able to access records, and others thinking that this

Fig. 5 Different options for how often care appointments (specialist and non-specialist) and coordination appointments take place (visual representation of taxonomy domain 4)

access should be limited (e.g., to necessary information such as the information relevant to the current care/condition, rather than all of the patient's history). Some patients highlighted that they would like to control who has access to this information. Workshop participants highlighted that full access to records with a summary may be helpful.

Domain 6. Mode of contact
Our findings highlighted different modes (see Table 4). Figure 6 provides a summary of this domain.

Information sharing/communication
For information sharing, participants described many different modes, including digital methods, written methods, verbal methods or a lack of information sharing.

Options for digital information sharing included digital records, letters, databases and registries (stored locally, in the cloud or on an app), portals (e.g. national online portals to access records, letters and guidance), mobile applications for patients (e.g. Patient Knows Best app or apps for patients to hold and share information from their records), and patient information. Digital options for communication between professionals (e.g. virtual panels, email hotlines, virtual meetings, email) and between professionals and patients (e.g. email, Whatsapp) were identified.

Many different written methods of information sharing were also identified: (1) written care documentation, (2) service planning, (3) guidelines and pathways. For written care documentation, this included: written records (e.g., condition specific passports and alert cards), letters, care plans (e.g. agreed care plans, education health and care (EHC) plans and transition plans) and reports. For service planning, written methods included plans to

specify hospital and professional roles and responsibilities, and standard operating procedures to record MDT working. For guidelines and pathways, this included evidence-based service specifications outlining service standards, quality assurance standards, national guidelines (e.g. NICE or charity produced), international best practice and training policies and frameworks for care coordinators and supervisors.

Telephone was also identified as a mode of communication between professionals (e.g. ringing others to coordinate care, conference calls, discussing treatment plans) and between professionals and patients (e.g. emergency point of contact and telephone advice services).

A lack of information sharing was also highlighted throughout the focus groups and interviews (including a lack of communication between team members and a lack of information sharing).

Care and coordination appointments
In terms of care and coordination appointments and communication, our participants described many different modes, including face to face, digital, phone and a combination of methods.

Face-to-face care delivery was identified for many aspects of care and coordination, including meetings (e.g., team meetings and patient/coordinator meetings), care appointments (e.g. initial patient/professional contact, at key treatment phases such as diagnosis and stabilisation, and for appointments requiring physical examinations) and support (e.g. peer support group meetings, network member meetings, and monitoring from charity nurses).

Digital options for care delivery and coordination appointments were identified. For coordinated care delivery, this included: video appointments with professionals

Fig. 6 Different options for mode of coordination (visual representation of taxonomy domain 6)
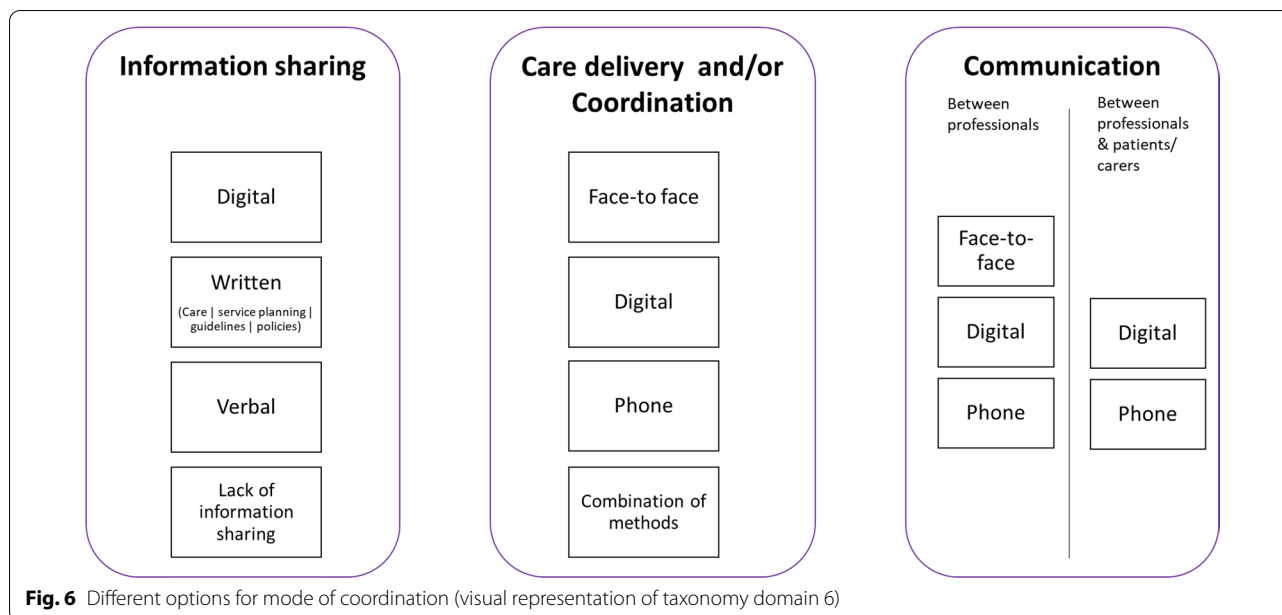
(e.g., Skype, Zoom, WhatsApp, Facetime), virtual centres and MDT clinics, digital monitoring (e.g., electronic wearable devices, apps to record test results), virtual tours of wards, diagnostic technology. For coordination this included: video appointments with coordinators (e.g., Skype, Zoom, Whatsapp, Facetime), coordination in the cloud, and virtual review (as the lowest level of coordination).

Options for telephone care delivery (e.g., clinics/consultations and catch ups) and coordination (e.g. calls with coordinators and introductions) were also discussed. Workshop findings highlighted that COVID-19 has accelerated the shift to digital and telephone delivery of care for people living with rare conditions.

Discussion
Key findings
We have developed a taxonomy of care coordination for rare conditions, based on learning from the UK health system and the National Health Service. We identified six domains of care coordination: (1) ways of organising care (local, hybrid, national); (2) ways of organising those involved in a patient's care (collaboration between many or all of those involved, collaboration between some of those involved, and a lack of collaborative working); (3) responsibility for coordination (administrative support, formal roles and responsibilities, supportive roles and no responsibility); (4) how often appointments and coordination take place (regular, on demand or hybrid); (5) access (full or filtered access to records), and (6) mode of information sharing, care coordination/delivery and communication.

How findings relate to previous research
These findings extend knowledge on care coordination. National policy documents and previous research have highlighted the importance of care coordination [3, 13, 32]. However, findings indicate that little is known about coordination for rare conditions and what this might entail [13]. Previous research has shown that coordination for rare and common chronic conditions has many components [12, 13, 33, 34], but care coordination had not been formally categorised. The taxonomy presented in this paper extends previous research by formalising care coordination for rare conditions into six domains (each with different options). Whilst previous research has developed taxonomies for related concepts [16–19], this is the first research that has attempted to develop a taxonomy of care coordination for rare conditions. Findings indicate that whilst different conditions have different characteristics and challenges, it is possible to develop one taxonomy that covers a range of conditions and a range of care coordination options.

Our findings highlighted three main options for organising care, including nationally centralised, hybrid and local care. This supports previous research which has highlighted the importance of specialist centres for people living with rare conditions [35] but extends previous research by demonstrating the potential usefulness of hub and spoke models and outreach models for rare conditions; models which are not new but which have been used in other chronic conditions with success [36–38]. Additionally, the findings highlight that local care is not necessarily problematic, with some participants articulating the role of local care and the benefits that this

provides them. However, findings do indicate that developing local expertise and knowledge is key.

Our findings on the organisation of professionals and the patient/carer for rare conditions supports previous research that indicates the importance of collaboration and MDTs for rare conditions [12, 39] and other conditions [40]. Findings also support previous research which indicates a need to join up care appointments from different disciplines and hospitals into one appointment (e.g., condition-specific clinics), in order to facilitate coordination [12, 13]. However, findings indicate that collaboration does not always happen in practice and that improvements in collaboration/joined-up working are needed. However, there have been some recent initiatives to improve collaboration across health and social care generally (e.g. the introduction of care coordinators in primary care networks).

Our findings extend previous research by outlining different types of responsibilities and roles needed to coordinate care for rare conditions. Previous research indicates the importance of care coordinators [12, 13, 41, 42]. However, findings extend this research by demonstrating the many different roles needed to coordinate care (administrative support, coordinators, clinical leads, GPs, charities and patients/carers). However, patient/carer involvement in coordination is not always appropriate if patients/carers are unable or do not want to coordinate their own care; and may have a negative impact on patients, families and the treatment burden that they experience [2, 12, 19, 39].

Our findings highlight the importance of following clinical guidelines and service specifications which outline how often appointments should take place. However, findings indicate that patient and provider factors need to also be taken into account when considering how often patients should be seen (see [20]).

Findings highlighted the potential for remote methods of coordination, including digital information sharing (e.g., through electronic records), virtual clinics and care coordination appointments. This shift to digital methods has been accelerated during the COVID-19 pandemic. This supports previous research, which indicates that digital methods may show some potential for use in health care delivery [43, 44]. Our findings suggest that this may also apply to care coordination, but that remote methods cannot fully replace face-to-face appointments.

Strengths and limitations

One strength of our study is that we used robust analysis procedures which strengthen the validity of this study. Twenty percent of data were coded by a second researcher. Additionally, the research team and members of the PPIAG were continually involved in discussions about analysis and findings. We also triangulated findings outputs from other parts of the CONCORD study [13, 21] to ensure no major omissions.

Whilst we sampled from a variety of rare conditions, locations and sectors, we were unable to include every rare condition. Some groups, including individuals from minority ethnic groups and certain roles (e.g. GPs) were under-represented. Therefore, whilst we collected extensive data, and included as many different views as possible, we are unlikely to have captured every possible option of care coordination. It is therefore possible that our taxonomy may not be applicable to all rare conditions, and there may be other domains or subdomains that we have not captured. However, we did speak with a range of patients, carers, professionals, commissioners and charity representatives with experience of a wide range of rare conditions and groups of rare conditions. To account for this possibility, we designed the taxonomy so that it can be applied flexibly (i.e. it is not expected that every care coordination option presented in this taxonomy would be appropriate for all rare conditions), and any application of this taxonomy should take into account a range of patient, professional, resource and societal factors that influence coordination, which will differ for all rare conditions (see [20] for factors influencing coordination and the development of hypothetical models of care coordination).

Additionally, the taxonomy was developed from data collected within the UK, and therefore it is likely that findings may only apply to the UK healthcare system. However, it is hoped that the taxonomy will also provide learnings in other healthcare contexts.

Previous research has demonstrated that one key challenge of coordination is that it is difficult to distinguish between aspects of care and coordination components [13, 45]. This is also a potential limitation of some options within our taxonomy (e.g., mode of care appointments, frequency of care appointments). However, we believe that the mode and frequency of such care appointments is part of care coordination, i.e., it may be that a care appointment that takes place virtually (with all health care professionals involved) may be more coordinated than other modes of care appointment (e.g., visiting different health care professionals for individual appointments). Additionally, this was not identified as a concern by any workshop participants.

The taxonomy is designed to cover health and social care received throughout a person's life; however, our sample may have been limited as many of the professionals included within our study were from a medical or paramedical background. Therefore, it is possible that these findings may not account for experiences and perceptions from some social care or mental health care providers not included in this study. However, many of

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the patient, carer and professional findings reflected on care coordination across both health and social care, in addition to other sectors such as education.

Implications

The development of the taxonomy could lead to the standardisation of terminology for care coordination in rare conditions. Previous research proposed that better measurement of systems for organising and delivering health care systems are needed [1]. This taxonomy will help to achieve this goal as it provides a better understanding of coordination and ways of organising and delivering health care for people affected by rare conditions. This will support researchers in operationalising and measuring care coordination. If care coordination strategies are piloted, evaluated and implemented more widely within the NHS, this may lead to better care and reduced burden for people living with rare conditions [9, 46].

The taxonomy can be used by health care professionals delivering care for people with rare conditions and as a menu for policy makers, service planners, researchers and commissioners to consider when developing new and/or existing models of coordination for rare conditions. For example, we have used the taxonomy, together with the findings on the factors influencing care coordination to develop some hypothetical models of care coordination that may be applicable in different situations (see [20]). These models take into account different situations such as whether the patient would like to be involved and can coordinate their care, where the patient and carer live, whether the rare condition has a specialist centre/service, and whether it is clear which professionals need to be involved in care. This is due to the large number of rare conditions and the need to ensure that findings can be tailored appropriately to different situations. To support appropriate tailoring for individual conditions, we developed a flow chart that may inform how these taxonomy findings, together with findings on the factors influencing care coordination can be used to develop such models (see [20]). These models can be costed and evaluated by researchers and services.

These findings could be particularly helpful during the development of the rare disease action plan in response to the new Rare Disease Framework [47]; in which care coordination is identified as a key priority. The taxonomy can also be used by researchers to evaluate models of care coordination. The taxonomy also provides clinicians and patients/carers with expectations about the different ways in which care can be coordinated. Findings can be used by clinicians to map the domains of care coordination onto their current services for rare conditions, but also to consider how services may adapt or develop their services for rare conditions in future.

Given similarities between common and rare chronic conditions that were highlighted in previous research [13], researchers interested in care coordination for other conditions may be able to adapt the taxonomy for use in other complex chronic conditions. Additionally, the process outlined in this manuscript could be adapted by researchers to develop comprehensive taxonomies to understand and organise other health care services.

Future research

Future research is needed to explore where different ways of coordinating care have been implemented and to evaluate the implementation, effectiveness, and cost-effectiveness of different models of care coordination for rare conditions in practice. This is important given that it is not yet clear whether coordinated care leads to better outcomes for patients/carers, professionals and organisations. Further research is needed to operationalise care coordination models so that delivery of care coordination can be measured.

Additionally, future research on care coordination and views on care coordination from professionals working in social care, mental health and other sectors such as education may be beneficial in determining whether the taxonomy can apply in these situations or whether amendments are needed.

Conclusions

Findings from our qualitative study with key stakeholders (patients, carers, health care professionals, charity representatives and commissioners) provide a thorough taxonomy of care coordination for rare conditions. Our taxonomy can facilitate the development and evaluation of existing and new models of care coordination for people living with rare conditions. The process outlined in this manuscript provides a template that could be adapted to develop taxonomies for other health care services.

Supplementary Information

The online version contains supplementary material available at https://doi.org/10.1186/s13023-022-02321-w.

Additional file 1: Topic guides for interviews and focus groups.

Additional file 2: Topic guide for workshops.

Additional file 3: Visual representation of workshop findings.

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Availability of data and materials
The datasets generated and/or analysed during the current study are not publicly available due to participant confidentiality but are available from the corresponding author on reasonable request.

Declarations

Ethical approval and consent to participate
This study received ethical approval from the London-Surrey Borders Research Ethics Committee of the Health Research Authority (reference: 19/LO/0250). Participants provided informed consent to participate in this study and for anonymised quotes to be published.

Consent for publication
Not applicable as all individual quotes are fully anonymised. As part of the consent to participate, all participants gave consent to their anonymised quotes being used in research publications.

Competing interests
HW, AIGR, EH, PLN, KLB, LB, JK, LK, MK, AGS, SM and NJF declare they have no competing interests. AS, AH and JJ are employees of Genetic Alliance UK. Genetic Alliance UK runs Rare Disease UK—a campaign for people with rare diseases and all who support them.

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