Co-ordinated care for people affected by rare diseases: the CONCORD mixed-methods study

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Scientific summary

The CONCORD study

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Scientific summary

Background

There are an estimated 6172 unique rare conditions. Each rare condition affects fewer than 1 in 2000 of the population, but combined they affect a large number of people, with more than 3.5 million people in the UK and 30 million people in the European Union affected by a rare condition. The problem addressed by this research project is the variation in how care is co-ordinated (and in many cases the complete lack of care co-ordination) for people affected by rare diseases in the UK. In this report, we use the terms 'rare conditions' and 'rare diseases' interchangeably to refer to rare, ultra-rare and undiagnosed diseases and conditions.

Rare diseases are often serious, chronic and complex in nature, affecting multiple systems of the body. As a result, patients often have several health-care professionals (HCPs) involved in their care. For many people, it is usual to have to access a number of different health services to receive the care they need, including care by specialists and care nearer to home. Care by specialists may require the patient to travel long distances and stay away from home, which can be inconvenient, costly and stressful. Care nearer to home may involve care by the local hospital or general practitioner (GP). Receiving care from a range of people, including specialists and local providers, can cause problems because co-ordination between the different professionals and services is often poor and care plans may not be in place or followed; as a result, some patients may experience gaps in their care because they do not see the right professionals and, when they do, the information the professional needs to facilitate appropriate care may not be to hand. The parents/carers of children with rare conditions often face a significant care burden, needing time off work to look after their children and take them to appointments. There can also be challenges in ensuring continuity of care when children transition from child to adult services.

There is some evidence to suggest that care is poorly co-ordinated for people affected by rare diseases. In addition, improving care co-ordination for people affected by rare diseases has been raised as a major concern by policy-makers, for example in *The UK Strategy for Rare Diseases* in 2013 [Department of Health and Social Care, Northern Ireland Executive, Scottish Government and The National Assembly for Wales. *The UK Strategy for Rare Diseases*. 2013. URL: www.gov.uk/government/uploads/system/uploads/attachment_data/file/260562/UK_Strategy_for_Rare_Diseases.pdf (accessed 5 October 2021)] and in *The UK Rare Diseases Framework* in 2021 [Department of Health and Social Care. *The UK Rare Diseases Framework*. URL: www.gov.uk/government/publications/uk-rare-diseases-framework/the-uk-rare-diseases-framework (accessed 17 January 2021)]. Unfortunately, although there are indications that care needs to be better co-ordinated for people affected by rare diseases, there is not good evidence as to how this should be achieved. The aims of this research project were to use quantitative and qualitative research methods to investigate (1) if, and how, care of people with rare diseases is co-ordinated in the UK and (2) if, and how, patients and families affected by rare diseases, and HCPs who treat rare diseases, would like care to be co-ordinated.

Objectives

We investigated the following five research questions (RQs):

1. What does 'co-ordinated care' mean, what are the components of co-ordinated care and in what ways, and why, may co-ordinated care for people with rare diseases be similar to or different from co-ordinated care for people with other conditions?

2. Is care for people with rare diseases in the UK co-ordinated and, if so, how?
3. What are the preferences of patients and families and HCPs in relation to how care for rare diseases is co-ordinated?
4. What are the different ways in which care for people with rare diseases might be co-ordinated?
5. How much do the different ways in which care for people with rare diseases might be co-ordinated cost?

Methods

For RQ1, we conducted a scoping review of reviews about care co-ordination for chronic conditions in general, and not just rare conditions, to identify factors important for co-ordinated care. This scoping review aimed to provide an updated definition of co-ordination of care for chronic conditions (both rare and common), to identify key components of care co-ordination for chronic conditions (both rare and common) and to explore whether or not the findings apply to rare conditions. We followed a recommended systematic approach to conducting scoping reviews. We undertook three focus groups to find out if the scoping review findings applied to rare conditions and to support the design of the survey and discrete choice experiment (DCE).

For RQs 2 and 3, we conducted a national cross-sectional survey of current experiences, incorporating a DCE of preferences for co-ordination. Survey participants were adult patients affected by a rare condition, parents/carers of children or adults with rare conditions and HCPs (e.g. doctors, nurses and allied health professionals) involved in the care of people with rare conditions. The content of the questionnaire was informed by 15 semistructured qualitative interviews with patients and carers to identify costs associated with living with rare conditions. These interviews were also used for an exploratory qualitative study of the impact on patients and carers of having care that was not co-ordinated.

For RQ4, we drew on the findings of the scoping review and also undertook 30 interviews and held four focus groups and two workshops with a range of stakeholders to develop and refine a taxonomy of different models describing how care for people with rare conditions could be co-ordinated.

For RQ5, we reviewed the costs of different components of co-ordinated care.

Study participants comprised patients (aged $\geq$ 18 years) affected by a rare condition, parents/carers (aged $\geq$ 18 years) of children or adults with rare conditions, HCPs (e.g. doctors, nurses and allied health professionals) involved in the care of people with rare conditions, national leads on specialist health-care commissioning, national patient groups and charities, and local providers and commissioners of co-ordinated care. Participants were accessed via patient and provider networks and organisations.

To meet our aims required substantial input from patients and families, in terms of both helping to design and participating in the research. The research team included representatives from a national charity that is an alliance of more than 180 patient organisations (Genetic Alliance UK, London, UK) and from national patient organisations with direct experience of living with rare conditions. This involvement ensured that patients' and families' priorities and needs were the focus of the study, and contributed to the design and management of the study, patient recruitment, data collection, interpretation of findings and dissemination. In addition, the members of the research team from these organisations also ran the study's Patient and Public Involvement Advisory Group, which involved managing and working with a group of six to eight patients and carers and meeting twice a year for the duration of the project. This group supported the development of resources and participant information, patient recruitment and dissemination of findings.

Results

Research question 1

Our scoping review included 154 review papers. Common chronic conditions were reviewed in 139 reviews, three reviews focused on a single rare condition and 12 reviews focused on both rare and common chronic conditions. Our new definition of co-ordinated care for rare conditions, which was derived from the scoping review, is as follows:

> *Co-ordination of care involves working together across multiple components and processes of care to enable everyone involved in a patient's care (including a team of health care professionals, the patient and/or carer and their family) to avoid duplication and achieve shared outcomes, throughout a person's whole life, across all parts of the health and care system, including: care from different health care services . . . care from different health care settings . . . care across multiple conditions or single conditions that affect multiple parts of the body, the movement from one service, or setting to another. Co-ordination of care should be family-centred, holistic (including a patient's medical, psychosocial, educational and vocational needs), evidence-based, with equal access to co-ordinated care irrespective of diagnosis, patient circumstances and geographical location.*
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Components of care for rare diseases that require co-ordination relate to administration, assessment and diagnosis, planning, review and evaluation, feedback, follow-up care, use of technology, support for patients, carers and families, and support for HCPs. Components that outline how care can be co-ordinated relate to someone taking responsibility, use of specialist centres/clinics, communication, support for patients and families and HCPs, multidisciplinary team (MDT) approaches, continuity of providers and development of care plans. Components that may influence or contextualise co-ordination are evidence-based practice (e.g. guideline-based treatment), individual differences in needs, wants and preferences, the wider health-care environment and access to treatment.

Many of the key components and issues for co-ordinated care apply to both rare and common chronic conditions. Important factors that may make it more difficult to co-ordinate care for rare conditions are difficulties in diagnosing rare conditions due to insufficient knowledge and ability to recognise symptoms, and a lack of condition-specific expertise due to small numbers of patients.

Research question 2

We found that care for people affected by rare diseases is not well co-ordinated, with patients having limited access to care co-ordinators, specialist centres and care plans. In our survey, only 12% of 760 patients affected by a rare disease had a formal care co-ordinator, and 14% of 446 parents/carers reported that the person they cared for had a formal care co-ordinator. Only 32% of patients and 33% of parents/carers attended a specialist centre for the rare condition. Ten per cent of patients reported having a care plan related to their rare condition, compared with 44% of parents/carers. Fifty-four per cent of patients and 33% of parents/carers had no access to a formal care co-ordinator, care plan or a specialist centre, with only 2% of patients and 5% of parents/carers reporting having access to all three elements.

Findings from our exploratory qualitative interview study of 15 patients affected by rare conditions and their carers revealed that lack of co-ordination resulted in delays/barriers to accessing care and a significant burden on patients and carers. These effects have negative impacts on patients' and carers' physical and mental health, as well as financial well-being.

Research question 3

We found that patients, parents/carers and HCPs all preferred better co-ordinated care. All three groups preferred services where the cost of attending appointments was lower, electronic health records were immediately accessible to staff, the lead consultant was a medical expert in the patient's specific medical condition, care was provided with the support of a care co-ordinator, a specialist centre was available and there was a documented emergency plan in place.

There were some differences between the preferences of patients and parents/carers and of HCPs. HCPs preferred that care was entirely co-ordinated on behalf of the patient by a care co-ordinator, whereas patients and parents/carers preferred that they decided how they wished to be supported by the care co-ordinator. In terms of emergency plans, all three groups preferred there to be a documented emergency plan in place, but the preferences of HCPs for this were stronger than those of patients and parents/carers.

Patients and parents/carers were hypothetically willing to pay £2509 for access to a specialist centre, £2470 for a consultant who was a medical expert in the patient's condition, £2442 for electronic health records that were immediately accessible to staff, £1367 for a documented emergency plan and £1306 for the support of a care co-ordinator.

Research question 4

We developed a taxonomy of care co-ordination for rare conditions that outlined the following six domains involved in co-ordinating care for rare conditions: (1) ways of organising care, (2) ways of organising teams, (3) responsibilities, (4) how often care appointments and co-ordination take place, (5) access to records and (6) mode of communication.

Ways of organising care ranged from local care provision where all care was delivered locally to care being delivered in national centres that serve all patients in the country with a particular rare condition. In addition, there were 'hybrid' options that combined both specialist and local care.

Ways of organising teams ranged from little collaboration (e.g. not having a MDT) to high levels of collaboration (e.g. all professionals working together to provide or discuss care in a condition-specific clinic or MDT meeting). Intermediate options included some HCPs working together (e.g. in joint clinics).

We identified different types of responsibility involved in co-ordinating care for rare conditions, including administrative, formal and supportive roles. Administrative support included help organising appointments and having a point of contact. Formal co-ordination responsibilities were those conducted by a co-ordinator, a clinical lead or a GP. Supportive roles were also identified, including those conducted by patients/carers and those conducted by charities.

Different time periods for care appointments and co-ordination activities included regular appointments, on-demand appointments and a hybrid approach that combines both regular care (at a minimum) and on-demand support.

Patients' and providers' access to records ranged from full to restricted.

Modes of communication related to information-sharing (e.g. digital/written/verbal), care delivery and/or co-ordination and communication (e.g face to face, digital, telephone) were identified. A range of different options were identified for each. Perceived factors influencing mode of information-sharing and care delivery included patient factors (e.g. age, condition and individual needs) and health-care environment factors (e.g. access to technology).

Research question 5

We used the taxonomy to develop some illustrative models of care co-ordination that may be applicable in different situations. We developed a flow chart that may inform how the findings are used to develop such models. There is a paucity of data on the costs of elements of co-ordinated care.

Conclusions

The findings of this study have two main implications. The first relates to whether or not care for rare conditions is co-ordinated. Our definition of care co-ordination for rare diseases takes into account the complexity of achieving co-ordinated care and the fact that several components of care need to be addressed to improve co-ordination. This definition serves as a useful guide for researchers, policy-makers and other stakeholders seeking to improve care co-ordination. Evidence of the lack of co-ordinated care for people affected by rare diseases is provided by our national survey, which found that, for the majority of people affected by rare diseases, care is not well co-ordinated. In particular, access to care co-ordinators, specialist centres and care plans is limited. The importance of the finding from our national survey was made clear by our exploratory qualitative interview study, which found that patients and carers are negatively affected by poorly co-ordinated care, in terms of their physical and mental health and their financial well-being. The importance of co-ordinated care was further strengthened by the findings of our taxonomy and our analysis of preferences, which showed that patients, parents/carers and HCPs all have a clear preference for better co-ordinated care.

The second implication relates to the ways in which care for people with rare diseases might be co-ordinated. Our definition of care co-ordination and description of the components of care co-ordination can be taken into account when considering how to improve co-ordination. The taxonomy developed in this study can be used as a menu for service planners, researchers and commissioners to consider when developing new and/or existing models of co-ordination. The qualifier findings from the taxonomy can also be used to inform decisions about which models of care co-ordination may be suitable for use in different situations, accounting for the preferences of stakeholders. This is particularly helpful, given the complexity of care pathways and service funding for rare conditions. We developed a flow chart that may inform how the findings from the taxonomy may be used to develop such models and their potential costs.

The main limitations of the study were that it was not possible to capture the experiences of people affected by every rare condition, our participant sampling may have been biased if study participants were systematically different from the population affected by rare conditions, our cost analysis was limited in scope given the paucity of available data and there is considerable uncertainty in the costs associated with different co-ordination models.

There is little evidence on the costs and benefits of different approaches to improving care co-ordination for people affected by rare diseases. Further research would be beneficial to develop feasible, clinically effective and cost-effective models of care co-ordination.

Study registration

This study is registered as NIHR Clinical Research Network Portfolio reference number 41132, Research Registry reference number research registry6351 and Integrated Research Application System reference number 254400.

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