White Paper

Shared Decision Making in Rare Disease in the United Kingdom

Contents

Foreword

There are always two experts in a consultation: the clinician and the patient. The clinician has medical knowledge and experience in evidence-based medicine and care. The patient is an expert in their own needs, values and personal circumstances – including experience of what it is like to live with their condition. Shared decision making (SDM) is a process of continual exchange of information between these two experts in order to build a trusting, collaborative relationship and a shared understanding of how best to manage the condition for this particular individual.

There are many benefits of SDM for patients.[1] With greater knowledge of the options available to them and the support to consider these based on their own preferences, patients feel better informed. Their confidence to participate also increases and they are clearer about what matters to them. Patients have more accurate expectations of the benefits and risks of treatments and, by increasing involvement in their care, adherence to treatment could be improved.

In the context of rare diseases, SDM is even more important as there is often not a gold-standard treatment or care option available. Clinician experience and patient knowledge may be limited, but patients certainly have invaluable experience of what it means to live with the condition, which should guide decision making. However, while SDM is promoted across all healthcare settings and embedded in healthcare policy, there is currently no specific guidance for clinicians and people living with rare diseases on why, when and how this model should be used and the strategies they could use to implement it.

Over the last few months, I have chaired a Working Group whose aim is to shine a light on the important subject of SDM in rare disease. Our collaboration has led to this white paper, which we hope presents a clear picture of the current barriers and needs within SDM in rare disease in the UK and opportunities for its implementation going forward. We hope this paper delivers a call to action for stakeholders in rare disease communities to facilitate the implementation of SDM. Ultimately, we want every patient with a rare disease to be able to say: "this is what matters most to me right now", and for them to receive care and support that is tailored to their own goals and preferences – care that values patients' lived experiences.

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The Working Group

In November 2020, a wide range of stakeholders with different backgrounds, experience and expertise came together virtually as the Working Group to discuss SDM in rare disease in the UK. The Working Group collaborated over several months to ensure all perspectives were considered in this white paper.

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What is shared decision making (SDM)?

SDM is a collaborative process whereby patients work together in equal partnership with healthcare professionals (HCPs) to make decisions about their care – including tests, treatments, management, information and support. SDM is essential when there is more than one treatment or management option – including the option to do nothing – and other decisions need to be made about care and support.

It is the responsibility of the HCP to share evidence-based information about the treatment and care options available, including potential benefits and risks, and to ensure that the patient has a good understanding of this information, sufficient to make an informed decision, should they wish to do so. At the same time, the HCP must seek to understand the values, needs and preferences of the patient and ensure that these are incorporated into the decision.

The patient should be supported to engage in the process and encouraged to share their current preferences, concerns and experiential knowledge of having a disease, and participate in SDM.

If the patient is unable to enter into SDM, then the patient's caregiver should be supported to take on this responsibility, sharing the patient's values, needs and preferences, to the best of their ability.

This mutual exchange of information, needs, understanding and acceptance of the decision is at the core of SDM.

The benefits of SDM in rare disease and the implementation challenge

The benefits of SDM in all healthcare situations and settings – including preventative, acute, chronic, palliative and end-of-life treatment and care in primary, community, hospital and mental healthcare settings – have been recognised for some time, with around 40 years of research and 15 years of policy supporting the process.[1-10] Well-informed and empowered patients, who have greater knowledge of the options available to them, are likely to feel increased confidence entering into dialogue with their HCP and are able to communicate their needs, values and preferences more clearly.[6,10] This allows them to make informed decisions that align with their own needs. With more accurate expectations of the benefits and risks of treatment options and greater involvement in their care, patient satisfaction and adherence to treatment is improved.[4,6,8,10]

Yet, even with all the known benefits and published policies, SDM is not routinely practised. Key barriers to routine implementation include a lack of understanding of its benefits, the perception that there is not enough time to practise SDM, HCP attitudes to patient involvement and patient-perceived power imbalances in healthcare consultations.[11-13] Additional challenges arise with SDM in rare disease due to the low prevalence of individual conditions which may result in less clinical management experience for the HCP and a lack of awareness for the patient. SDM in rare disease might be considered unnecessary due to limited or non-existent care or treatment options or effective diagnostics.

However, in the context of rare disease, SDM is very important, because there is often no gold-standard treatment or care option available and the decisions will be preference sensitive. With limited or even no treatment options for some rare diseases, the best decision for the patient will depend on their individual needs and preferences. Therefore, it is vital to incorporate the patient's values and preferences in the decision-making process to improve the quality of their treatment and care.

To encourage SDM in rare disease, an understanding of the barriers and unmet needs for its routine implementation is first needed.

Barriers to SDM in rare disease

Healthcare professional approaches

Traditionally, a paternalistic approach has been adopted by HCPs, as they believe that, with their medical knowledge and experience, they are able to make the right decision on behalf of their patients.[10] Patients often expect this approach and may feel reassured by a 'supervisory relationship' providing them with a feeling of protection.[10] However, by understanding the patient's particular needs and preferences, the HCP is likely to learn valuable information, which would help them to adapt the suggested care and support plan for their patient. This understanding would more likely lead to decisions that are right for the individual patient and may, in turn, help the patient to better understand and adhere to their treatment.

For successful SDM, HCPs need to encourage patient engagement in the decision making process, share information about the disease and possible care and treatment options, seek information from the patient on their needs, values and preferences, and ensure that these are incorporated into the final decision that is made regarding care. At the same time, HCPs need to reassure patients that they are not alone in making decisions about their care and that it is a collaborative process.

RECOMMENDATION 1: HCPs seeing patients with rare diseases should receive skills training in SDM to understand the value of this process, how it differs from other forms of communication and how it can be enacted in routine clinical settings to support and engage patients with rare diseases.

Patient knowledge, attitude and confidence

When a patient (or their caregiver) receives the initial diagnosis of a rare disease, their knowledge of the condition may be very low, they may feel overwhelmed and they may lack confidence to enter into a meaningful discussion with the HCP. A power imbalance exists, whereby the patient or caregiver views the HCP as the expert and so places their trust in them to make the best possible decision about their care.[14] It is likely that patient-perceived power imbalances in rare diseases are exacerbated, when compared to other diseases, due to the highly-specialised and rare nature of these conditions. Furthermore, learning difficulties or lower levels of education can be a barrier to improving the patient's knowledge about their condition. Additional challenges to an effective SDM process exist when there are language and cultural barriers to communication. Cultural issues, such as mistrust of health authorities, can lead to lower than expected patient numbers for rare diseases which are genetically linked to particular ethnicities, while cultural discordance can be a barrier to SDM.

However, the patient may not appreciate that they themselves are experts with complementary knowledge about their own needs and lived experiences, and are therefore equal partners with the HCP in the SDM process. They may also feel that there is an expectation for them to be passive and compliant with the expert's opinions or they may be concerned that, by asking questions, they will add to the HCPs workload. This attitude can create a further barrier to SDM.[10,14]

Although uncommon, patients or their caregivers may sometimes come to a correct diagnosis of the rare condition themselves through internet research, after years with no formal diagnosis. At a later stage of the journey many patients or their caregivers are well-informed while generalist HCPs are less-informed.

Patient and caregiver knowledge, attitude and confidence can be improved over time if the HCP:

- provides accessible disease information, including self-care and chronic disease management, which is targeted to the individual needs of the patient;
- explains the rationale for patient involvement and the importance of understanding their preferences;
- describes the SDM process; and,
- explicitly encourages and supports the patient or their caregiver to participate.

However, information to increase patient and caregiver knowledge – which is targeted to their current needs – is not always available. For some rare diseases, patient information is extremely scarce and so engagement with patient organisations is vital.

RECOMMENDATION 2: A range of patient materials, including disease information factsheets and treatment option information (where available), an explanation of the SDM process and rationale for patient/caregiver involvement should be developed to meet the different needs of patients and address any barriers to communication, such as language and culture.

Changing needs during the patient journey

As a patient continues on their journey after their initial diagnosis, they become more informed about their disease and the questions they wish to ask, and their confidence is likely to increase. At some point the patient (or their caregiver) may become an expert in the condition – depending on the volume of information available to them – and therefore feel more confident to actively engage with SDM.[10]

If the rare disease is genetic, the patient or caregiver may already be relatively knowledgeable about the condition at the time of diagnosis due to member(s) of their family already having a diagnosis. As the patient continues on their journey, their experiential knowledge of the rare disease grows. They will start to know more about how the disease affects them than their HCP, which is an important element to feed into the SDM process.

During the patient's journey, it is not just knowledge, experience and confidence that change over time. The patient may also become an adult or experience changes in their personal life, such as in employment, finances, lifestyle, relationships and support network. These changes can impact their care needs and preferences, which, in turn, will influence their treatment choices. It is therefore vital that patients continue to share information regarding their current needs and preferences with HCPs and that HCPs encourage this information exchange on an ongoing basis.

RECOMMENDATION 3: A holistic and ongoing needs assessment – which can then be linked to the patient's care and support plan – should be conducted to increase the HCP's understanding of where the patient is on their journey, their individual goals, needs and preferences, and changes in these over time.

Distributed and co-ordinated care

The nature of a rare disease means that the time taken to receive the correct diagnosis may have been significant. The patient or their caregiver may therefore feel overwhelmed during their first appointment with a specialist HCP. Typically, multiple HCPs with different disciplines in various clinical settings might be involved in rare disease care/management if the condition impacts more than one part of the body, which can make co-ordination of care a challenge. Effective SDM relies on a shared understanding of the patient's goals, needs and preferences from all HCPs involved and potentially a distribution of the SDM tasks if the HCPs are from the same team. SDM in rare disease is therefore extremely valuable when multidisciplinary and multispecialty teams are involved to help the patient feel empowered and in control of their care. If a care co-ordinator has been appointed to take overall responsibility for the patient's clinical care, they are ideally placed to facilitate the SDM process across teams.

RECOMMENDATION 4: Training in SDM in rare disease for members of multidisciplinary and multispecialty teams and care co-ordinators is required, to support shared responsibility for SDM.

Effective and timely communication

Effective communication lays the foundation for a trusting relationship and this is essential for patient or caregiver engagement in SDM.[10,15] HCPs can help to promote effective communication, especially soon after diagnosis, by ensuring it is tailored for the patient or caregiver as medical terminology used by the HCP may be confusing for them if they are not knowledgeable about the condition.[10] Patient engagement can be supported by the HCP encouraging the patient to share their personal needs and preferences to feed into the SDM process.[11]

Time constraints of scheduled appointments may discourage the patient or caregiver from sharing information or asking questions, preventing development of a trust-based HCP-patient relationship.[10] Providing a clear explanation of the aim of the consultation, treatment/care options and what is expected of the patient/caregiver in the SDM process could help HCPs use the time more effectively. Ultimately, face-to-face appointments may not be the optimal time or place for patients or their caregiver to consider options or make decisions. Therefore, pre- and post-consultation communication is essential, possibly via virtual meetings, phone calls and emails.

Communication within a multidisciplinary/multispecialty HCP team and between the patient or caregiver and the team can be an additional challenge. All team members share responsibility for effective communication and need to contribute to SDM as each member will have a different experience with, and may gain different insights from, the patient.[11] A care co-ordinator can assist in effective communication in this situation, coaching both the patient/caregiver and team in the SDM process.

RECOMMENDATION 5: To support patients/caregivers to successfully engage in SDM, effective communication is needed: a pre-consultation holistic needs assessment that feeds into the consultation, supportive and preference-focused communication during the consultation and a post-consultation follow-up to understand patient-reported outcomes/experiences. This is particularly important if the patient is not allowed to be accompanied in the consultation due to hospital attendance restrictions such as those seen during the COVID-19 pandemic.

Integration of SDM with current practices and procedures

HCPs may feel that they already involve their patients in decisions and that an alternative process is unnecessary and will demand too much of their time.[11] However, evidence suggests that not all HCPs communicate the benefits and risks of treatment/care options or feel they have the time to gain an understanding of the patient's needs, values and life experience[11] – both of which are required for effective SDM.

To make SDM routine practice, health organisations (for example healthcare practices, hospitals or networks of clinics) must understand the value of this process, what it entails and how it differs from their current procedures and the process of informed decision-making.[11] SDM can then eventually be integrated into the organisation's current practices effectively to add value without taking significant additional time or resources.

Organisational support and leadership that encourages the use of SDM as the approved method of decision-making is critical for full implementation of this process in order to drive engagement from HCPs.[11,15]

RECOMMENDATION 6: Awareness of the benefits of SDM should be raised in health organisations and leadership teams to encourage engagement in SDM and to facilitate the integration of SDM into current practices.

Unmet needs

Patient and caregiver empowerment and engagement

Patients and caregivers are only able to fully engage in SDM if they feel empowered to do so.[16] In the field of rare diseases, patient/caregiver empowerment is even more valuable due to low prevalence of the disease and a resultant insufficiency of medical expertise and information. Increasing a patient's/caregiver's knowledge of the disease is a key part of facilitating empowerment.[16] However, knowledge alone is not enough and patients/caregivers also need explicit encouragement to feel 'permitted' to ask questions, to raise their concerns and to share preferences.[14,17] Ideally, this should be a two-stage process where patients/caregivers are 'prepared' to engage before a consultation and then 'prompted' to engage during the consultation.[18]

Patient and caregiver empowerment improves the collaborative relationship with HCPs, making patients/caregivers more likely to enter into a dialogue and engage in the decision-making process.[19]

RECOMMENDATION 7: To empower patients and caregivers, disease information materials explaining the SDM process and their role as an expert and a holistic needs assessment should be given to the patient/caregiver prior to the consultation ('preparing' the patient/caregiver). The patient/caregiver should then receive explicit encouragement to share their goals, needs and preferences during the consultation ('prompting' the patient/caregiver). This is particularly important if the patient/caregiver is not allowed to be accompanied in the consultation.

Psychological care

Rare disease patients can experience increased anxiety, stress and psychological challenges due to the length of time to diagnosis, difficulty coming to terms with this diagnosis and the reality of a chronic condition.[20,21] This can be compounded by feelings of guilt associated with hereditary diseases and complex family dynamics, which may influence the patient or their caregiver's decision-making capacity.

Psychological challenges can hinder the patient's or their caregiver's ability to assimilate the information provided, leading to difficulty with engaging in SDM and potentially unrealistic expectations of the treatment.

RECOMMENDATION 8: Psychological/emotional support should be integrated into the SDM process, with patient information and HCP communication training addressing how to engage in SDM in the context of psychological challenges.

Paediatric and neonatal care

SDM should be an essential part of managing all children with rare diseases, regardless of whether they have a treatable condition with a good long-term prognosis, a chronic disabling condition with lifelong healthcare needs, or a severe life-limiting condition. SDM is particularly relevant for children going into palliative care, where end-of-life decisions need to be made jointly between patients, parents and HCPs.

Duty of care is an important part of decision-making in paediatrics, as not all children have the capacity to understand or consent to medical treatment and so their parents or caregivers, together with HCPs, are required to make decisions on their behalf. A difference in opinion between the parents or caregivers and HCP regarding the best course of action for the child creates challenges for SDM. Furthermore, SDM in acute situations in paediatric and neonatal practice, when urgent interventions are needed, can be especially difficult.

RECOMMENDATION 9: Guidance on SDM in paediatric and neonatal care should be provided to parents/caregivers at the earliest opportunity, along with the holistic needs assessment so that the patient's family can be supported to consider their child's preferences and to ensure that the HCP understands the preferences and values of the patient's family.

Transition from paediatric to adult care

Moving from paediatric to adult care can be a particularly challenging time, as patients experience a transition from potentially a more paternalistic approach to their clinical management, to one where they are increasingly encouraged to share in the decision-making process.

To aid transition, patients may require a more supported period of shared responsibility. However, some patients may never have the capacity to independently engage in SDM without the continued support of their parents or caregivers due to learning difficulties. Furthermore, specialist HCPs in adult care may not be as experienced in conditions where, historically, children did not survive into adulthood.

RECOMMENDATION 10: Patient materials focusing on SDM in rare disease should be developed specifically for patients and their parents/caregivers for the transition from paediatric to adult care.

Opportunities

Virtual communication and consultations

During the COVID-19 pandemic, the use of virtual communication and consultations has increased. For many, this has been a positive development, increasing access and convenience, and possibly helping to reduce the perceived power imbalance between the HCP and patient, as, for example, the patient is not the 'guest' in the HCP's office, leading to a more equal conversation. Remote services and the ability to text, email and send photos, has helped to flatten the authority gradient leading to a more inclusive conversation.

For younger patients, however, virtual consultations may be challenging. HCPs may miss non-verbal cues and physical signs in virtual communications and consultations that would otherwise have been noticed during face-to-face appointments – in particular with regards to the interaction of young children and their families or caregivers. This creates the potential for HCPs to miss safeguarding issues for young patients. Conversely, a virtual consultation may allow a private one-to-one conversation without the dominant influence of the parent or caregiver and might improve patient disclosure.

Collaboration with patient organisations and peer support

For people with rare diseases, it can be a challenge to meet peers due to the rarity of their conditions. Patient and carer organisations are therefore extremely important to enable people with rare diseases to share experiences with other people with the same condition.

Patient organisations may act as advocates for SDM by promoting the approach amongst the rare disease community, by explaining that the patient's goals, needs and preferences are important and that these should be shared with the clinical team and therefore increasing patient/caregiver empowerment. Furthermore, patient organisations may provide patient information materials and support patients during consultations where possible.

UK Rare Disease Framework

The UK Rare Disease Framework was published in January 2021. This framework follows the 2013 Department of Health & Social Care UK Strategy for Rare Diseases, which ended in 2020. An opportunity exists to include SDM in the action plans that will be prepared based on this framework.

NICE SDM guideline development

New guidelines on SDM in adults are currently being developed by NICE, which focus on SDM as part of everyday care in all healthcare settings and promote ways for HCPs and patients to work together to make decisions about treatment and care. The guidelines makes recommendations on training, communicating risks, benefits and consequences, using decision aids and how to embed shared decision making in organisational culture and practices. The guidelines do not mention SDM in rare disease specifically; however, it may be beneficial to include this topic in the guidelines. The consultation for the guidelines is scheduled to take place in the first quarter of 2021.

Conclusion and recommendations

SDM in rare disease is beneficial for patient experience, outcomes and wellbeing, yet not often implemented. It is essential that both HCPs and patients/caregivers are provided with the skills, knowledge and tools required in order to take advantage of the valuable approach of SDM.

With increased awareness of the importance of SDM, particularly in rare disease, patients/caregivers will feel more empowered and will therefore be more likely to engage in SDM. Furthermore, HCPs will see the value in taking a patient's needs and preferences into consideration for an optimal decision-making process, without feeling like their medical expertise is questioned.

This white paper contains a number of recommendations for how to address HCP and patient culture, attitudes, approaches and other barriers and unmet needs for the successful implementation of SDM in rare disease. Effective and timely communication is a cornerstone of most recommendations, with communication pre- and post-consultation being essential.

Pre-consultation:

- The patient/caregiver should be supported and encouraged to complete a holistic needs assessment.
- Information on the rare disease, targeted to the patient, should be provided.
- The patient/caregiver should be given an explanation of SDM and their role in this process.

During the consultation:

- An explanation of the benefits and risks of treatment and care options should be given by the HCP, in a way that the patient/caregiver can understand.
- The HCP should elicit information from the patient regarding their needs, values and preferences.
- A discussion should take place on the options that are preferred by the patient, with the possibility of delaying the decision until the patient/caregiver has had time to reflect on the options.

Post-consultation:

- The patient/caregiver should be given an opportunity to ask any additional questions, engage in further dialogue and make or change decisions.

The support of health organisations will help integrate SDM into routine practice by taking a top-down approach to encourage the use of SDM at the HCP level. Training in the benefits and practice of SDM, including the relevant approach from HCPs, will aid the success of this integration.

This white paper is a call to action for stakeholders working in the rare disease sector to raise these recommendations within their communities with a view to their implementation in the real world.

Practical responses could include:

- Production of video clips for patients – by patients – explaining SDM, how it can be implemented, its benefits and the role of patients/caregivers and HCPs in the process.
- The development of disease-specific toolkits and holistic needs assessment templates that are co-developed by patient organisations and service providers for a specific rare disease community.
- Examples of pathways of care integrating SDM at each step.
- SDM skills training programmes for health organisations and HCPs.
- Clinical, managerial and patient organisation leadership for SDM at the national, regional and local level.

By taking action together, we can make a positive difference to the lives of those living with a rare disease.

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