Patient Involvement in Rare Disease Decisions: What Makes Participation Meaningful?

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Rare Disease Watch | November 2026
White paper | Version 1.0
Published on https://rarediseasewatch.com/

Executive summary

The cystic fibrosis community has revisited its research priorities as treatments have changed. Patients, families and healthcare professionals helped choose the questions, then returned to them several years later. [1,2] That published example offers a useful starting point for a wider question: when people contribute to a rare disease decision, how can they see what happened to their views?

The evidence is encouraging in places and frustratingly incomplete in others. Rare disease reviews describe contributions to research questions, study design and the choice of outcomes, but find much less consistent assessment of their effects. [3,4] Being able to describe an activity is easier than establishing what difference it made.

This paper examines involvement in research, service development and governance. It distinguishes consultation, where an organisation seeks advice but retains the decision, from arrangements in which patients and carers share authority. Both can be useful if people know what they are being invited to do. The difficulty comes when the language promises more influence than the role allows, or when nobody returns to explain the decision.

The practical proposal is to keep an accessible record of recommendations, responses and subsequent action. Alongside that record, organisations need to consider who could take part, whose experience was missing and what participation cost people in time, money or energy. A change on paper tells only part of the story.

This is a targeted narrative synthesis of selected research and guidance available by 1 November 2026, rather than a systematic review. Its practical proposals are recommendations for discussion, not a validated assessment tool. It makes no claims about Rare Disease Watch outcomes and includes no patient testimony.

Scope and evidence selection

This paper concerns the decisions people help make about research and services. It covers research priorities, study design, service development and governance, rather than individual treatment choices. Taking part in a trial is a different role from helping design it; someone can do both, but one does not automatically confer the other.

Targeted web searches were undertaken on 1 November 2026, combining rare disease with terms including patient involvement, research priorities, impact, evaluation, governance, accessibility and payment. Sources were selected for their relevance to rare diseases, the transparency of their methods or their role as institutional guidance. Central claims and reference details were checked against journal, bibliographic or issuing-organisation pages. Where full text was not readily accessible, some checks relied on abstracts.

The selected sources include two rare disease reviews, a priority-setting partnership and its later refresh, evaluation and reporting resources, and institutional guidance. They answer different kinds of questions. A study can describe what happened; a standard can set out what ought to happen. Agreement about good practice is not evidence that it has been achieved.

The search was purposive and limited to English-language sources. It did not include exhaustive database searching, duplicate screening, formal assessment of bias or statistical pooling. UK and European sources predominate, and the older reviews describe the evidence available at their own cut-off dates. These limits matter when applying the discussion elsewhere.

Terms used in this paper

Consultation means asking for views while retaining the decision. Shared decision-making, as used here, gives contributors an agreed part in choosing between options. Co-production suggests working together across the relevant stages, which asks more of an organisation than including patients in a workshop.

Demonstrable influence is a separate question: can a contribution be connected to a change in an option, process, decision or its reasoning? These are working distinctions for this paper, rather than a universal ranking of involvement methods.

The appropriate arrangement depends on the work. Checking whether a leaflet is understandable may need a short consultation. Choosing a research agenda needs room to question which problems deserve attention in the first place.

What the research can show

Forsythe and colleagues' 2014 review included 35 studies. Patients and other contributors helped with research agendas, outcomes, design and recruitment, while rare disease organisations also connected communities with researchers and supported infrastructure. Reported benefits were not matched by empirical assessments of engagement practices and their effectiveness. [3]

The review could therefore describe contributions more confidently than it could compare their effects.

A similar difficulty appears in Velvin and colleagues' 2022 study. Their scoping review included 13 articles, alongside a survey of staff in Norwegian rare disease centres. Researchers described potential gains in quality and relevance, but methods for assessing impact remained limited. [4] Their accounts help explain why involvement is valued and where it is difficult; they cannot establish that it caused better research outcomes.

Research priorities in cystic fibrosis

The cystic fibrosis James Lind Alliance partnership gives a more concrete view of what joint work can produce. People with the condition, families and healthcare professionals helped identify and prioritise unanswered treatment questions, producing a top ten published in 2018. [1] The questions themselves are an output that others can examine.

The priorities were refreshed in 2023 as the treatment context changed. The authors reported that the earlier work had informed substantial research funding. [2] This suggests a connection between the questions chosen and later research activity, although it cannot tell us what would have been funded without the partnership. Nor does funding alignment, on its own, establish better health outcomes.

Patient input into study design and outcome selection is described in the rare disease literature. [3] To understand an individual study, it helps to see the documents: what was proposed before involvement and what was altered afterwards? An added outcome, a changed appointment schedule or a clearer explanation of risk gives the reader something specific to assess. The phrase "more patient-centred" does much less work without that detail.

A revised protocol can show a design change. Whether recruitment improved or participants found the study less burdensome needs its own assessment, taking account of other influences such as eligibility, transport and disease progression.

Whose experience enters the decision

Someone who travels regularly to a specialist centre may see different problems from someone who has never managed to reach it. Both experiences belong in a discussion about access. A small rare disease population can still contain considerable differences in age, disease stage, language, treatment options and family circumstances. Small numbers do not make those differences disappear.

The UK Standards for Public Involvement offer a useful frame for thinking about this. They cover inclusive opportunities, working together, support and learning, governance, communications and impact. [5] Their purpose is reflection and improvement; they do not provide a score that settles whether a particular group is representative.

It helps to say how someone's contribution should be understood. Are they describing their own experience, bringing views gathered from a network or speaking through an appointed organisational role? Each can add something, but a personal account should not quietly become the reported position of an entire community.

One person's experience can matter without standing for everyone's.

Patient organisations can be valuable routes into involvement. Where feasible, clinicians, community services and accessible public invitations may reach people outside those networks. Statistical representativeness may be unrealistic in a very small population, but an organisation can still explain whom it hoped to reach, who contributed and where gaps remain. That account needs enough detail to be useful without making individuals identifiable.

Patients carers and experienced advocates

Carers bring knowledge of coordination, daily support and family burden. Their perspectives may overlap with those of the person receiving care, or differ from them. The distinction deserves to remain visible. Before assuming that somebody needs another person to speak for them, consider accessible communication, support and age-appropriate ways to contribute. These are practical recommendations from this synthesis.

Understanding technical language should not disqualify an experienced advocate. It may be precisely what enables them to challenge a proposal. Organisations can value that experience while helping newer contributors join through induction, paired roles or alternatives to speaking in a meeting.

There will also be disagreement. A minority view may expose a trade-off that a final ranking conceals, especially if a decision suits one group while leaving another with an unresolved problem.

Accessibility and payment

An online meeting may save someone a long journey and still be difficult to manage. Fatigue, communication needs, unreliable internet or caring responsibilities can affect whether a person can contribute, and for how long. The recommendation here is to discuss access before choosing the format and return to the question as the work develops.

Shorter meetings, breaks, advance papers in plain English, captions, interpretation and telephone access are possible starting points. Written or recorded responses can give people time to think outside a live discussion. But an alternative format is only useful if the contribution reaches the decision-makers. Contributors need to know how that will happen, just as they need documents with clear headings, readable tables and meaningful links.

The chair has a part to play too, by explaining unfamiliar terms, making room for responses and avoiding pressure to disclose personal information. Someone should be able to decline a question or pause their involvement without having to defend the decision.

Payment and the cost of contributing

Health Research Authority guidance recognises payment as a way to acknowledge contributors' time and experience. It distinguishes payment from reimbursement and highlights questions about employment status, tax and welfare benefits. [6] Those are UK-specific considerations; arrangements elsewhere need local advice.

For the work considered here, I recommend budgeting for preparation and follow-up as well as meetings, alongside agreed access and caring costs. Explain the rate, the claims process and when payment should arrive. Where possible, arrange necessary expenses directly or in advance. Otherwise, an invitation may depend on someone being able to cover costs while waiting for reimbursement.

People should also be free to decline payment, without their choice becoming the expectation for others.

Payment recognises work. It should leave people free to criticise the organisation or disagree with its preferred option. It does not, by itself, tell us how much authority a contributor holds: a paid advisory role may still carry no vote.

The evidence selected here does not establish an ideal rate or meeting format across rare diseases. A useful evaluation would ask contributors what participation required of them, and whether the arrangements remained manageable over time.

Service development and governance

Service decisions raise some of the same questions as research, but research findings cannot simply be carried across. NHS England's specialised commissioning framework describes routes for patient and public involvement in commissioning and governance. [7] It establishes an expectation of participation. To understand how that works locally, we still need to examine the roles and decisions themselves.

A written role description should make clear whether patient members can propose agenda items, request information, vote, seek reconsideration or record dissent. It should also identify who makes the final decision. An advisory seat may be useful, but describing it as shared control gives contributors and the public a different impression.

The European Medicines Agency distinguishes community representatives, organisational representatives and individual experts, and describes involvement across the medicines lifecycle. Its guidance also requires compliance with competing-interest policies. [8] These distinctions help explain the capacity in which someone contributes; they do not imply that each role carries the same authority.

Decisions about services

Consider a proposed service redesign. Patients and carers could help examine referral routes, travel, appointment coordination and communication between services. This is a suggested application, not a Rare Disease Watch case study. If the service model has already been settled, asking contributors to help explain it leaves a much narrower role than inviting them to question how it should work.

Some limits will be real. Staffing, safety or funding may constrain the options. Explain the constraint and allow people to question the account where possible. If a recommendation is declined, a specific reason lets contributors judge whether an alternative was considered; a general reference to "feasibility" often leaves that unclear.

The approach proposed here includes disclosure of relevant organisational funding and individual interests, with proportionate rules applied consistently. Contributors also need somewhere to raise a concern outside the relationship with the chair. This is particularly relevant when the organisation inviting involvement is connected to their care or access to opportunities.

Follow-through needs an owner. If a contributor becomes unwell or leaves, the organisation should still know what it agreed to do and who is responsible for doing it.

Evaluating influence on decisions

Vat and colleagues developed a framework for evaluating patient engagement through a participatory process that included application in 24 cases. It separates resources and activities from learning, changes and wider impacts, allowing measures to be chosen for the context. [9] That helps organisations decide what to examine, though it does not establish that using the framework improves patient outcomes.

GRIPP2 addresses reporting. Its short checklist covers the purpose, methods, results, outcomes and critical reflection on patient and public involvement. [10] Clear reporting gives readers a better chance of assessing the work. Whether power was shared or a decision improved still requires examination.

Recording recommendations and responses

I propose the record below as a practical starting point, informed by those resources. It is not a validated score or accreditation scheme. An internal version can hold the detail, with an accessible summary shared in a form that protects contributors' privacy.

Record	Question it should answer
Decision and starting position	What was open to change, and what was originally proposed?
Contribution and context	What was recommended, by whom in which role, and which perspectives were absent?
Response and reasoning	Was it adopted, modified, deferred or declined, and why?
Implementation and review	Who will act, by when, and how will completion be checked?
Contributor feedback	Do contributors recognise this account, and what remains disputed?

Suppose a trial advisory group recommends fewer hospital visits because travel is burdensome. This hypothetical example could be recorded by showing the original schedule, what was revised and any safety constraints. A later assessment could ask participants about the burden. Improved recruitment would be a separate claim requiring separate evidence.

Attendance and satisfaction tell us something about the process, but they do not settle the question of influence. Even the proportion of recommendations accepted can mislead if many small changes obscure a central concern that was never discussed. Conversely, a decision to keep the original option may still reflect careful consideration of patient input. The record should make that reasoning available for examination.

Implications and unresolved questions

Start with a decision that is still open, an owner and a date after which options will close. Agree what contributors can influence before asking for their time. Recruitment, access and payment then become part of preparing the work, rather than problems to resolve after invitations have gone out. These are recommendations from this synthesis, not a tested package of interventions.

Keep the record as the work proceeds. Include contributions made outside meetings, recommendations that were declined and matters still unresolved. When reporting back, give people a chance to correct the organisation's account. That may reveal a disagreement about what was heard, even where the final decision has not changed.

For researchers, this means connecting involvement to protocol and outcome choices. For patient organisations, it includes being clear about the basis on which views are offered. Service leaders and policymakers need to specify authority and follow-through. A prospective contributor should be able to understand the work and its limits before deciding to take part.

What remains uncertain

The selected evidence cannot establish a best model for rare diseases or compare the effectiveness of recruitment, payment and governance arrangements with confidence. Reporting and measurement weaknesses persist in the rare disease reviews, while evaluation frameworks help organise questions without resolving causation. [3,4,9,10] There may also be more to learn from unsuccessful or abandoned partnerships than published accounts currently reveal.

Several people may contribute to a change at once. A decision record can show a credible patient contribution without proving that it was the sole cause. Stronger claims would need prospective evaluation, comparison where feasible and independent review. Such work should examine burden and exclusion as well as the outcomes organisations hoped to achieve.

There are limits to whose experience this paper can illuminate. It does not offer a comprehensive synthesis of lower-resource settings, involvement by children or people with communication disabilities, or people outside established patient organisations. Further work needs to seek those perspectives, rather than assume that existing guidance adequately captures them.

An organisation may listen carefully and still reach a decision some contributors dislike. Agreement cannot be the only test. What matters is whether people can recognise how their experience entered the reasoning, understand what followed and question an account that leaves something out.

When the decision is read back, can they see the work they helped to do?

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