

Child Rights and Wellbeing Impact Assessment (CRWIA)

Progress Report on Scotland's Action Plan for Rare Diseases (2024)

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Disclaimer

This **draft** document is an **initial assessment** of the impact of our first Progress Report on Scotland's Action Plan for Rare Diseases (2024). Scottish Government will continue to review and update this document where required during the strategic decision-making process. Any future iterations will reflect an increased understanding of these impacts as the amount of data and research available continues to grow.

This impact assessment should be read in conjunction with the Equality Impact Assessment and the Fairer Scotland Duty Assessment.

Child Rights and Wellbeing Impact Assessment Template

Introduction

1. Brief Summary

Type of proposal (Please state):

Bill No Scottish Statutory Instrument No

Decision of a strategic nature relating to the rights and wellbeing of children Yes

Progress Report on Action Plan for Rare Diseases (2024)

Key Terms explained

chronic – something bad (such as a disease) continuing for a long time

diagnostic odyssey – the time between when a symptom or feature of a genetic or rare disease is noted to the time when a final diagnosis is made

geneticist - someone who specializes in the science of genetics, which is a branch of biology that deals with the characteristics of different organisms (something that makes up a living thing). They look at the development of such characteristics, and how these characteristics are passed on

A rare condition is defined as a condition which affects less than 1 in 2,000 people. Approximately 412,080 people out of a population of 5,151,000 in Scotland have a rare condition, and it is estimated that there are over 7,000 different rare conditions. Rare conditions are thus individually rare, but collectively common, with 1 in 17 people being affected by a rare condition at some point in their lives. Most rare conditions are chronic and will require management throughout a person's life.

The UK Rare Diseases Framework, co-produced by the UK Government and devolved nations, was published in January 2021. This identified four shared priorities, following extensive stakeholder engagement, to improve the lives of people living with a rare condition: faster diagnosis, increased awareness among healthcare professionals, better co-ordination of care and improved access to specialist care, treatments and drugs. Working to these four priorities, each administration's task was to publish a nation-specific Action Plan by the end of 2022. Scotland's Action Plan for Rare Disease was published in December 2022. We are now publishing our first report on our progress implementing our Action Plan. Therefore the parameters of this CRWIA will focus on the impact of this progress.

Our Action Plan was written on the principle of 'What matters to you', foregrounding the feedback we had received from community engagement. This Progress Report will continue giving the same importance to what people and families living with a rare condition consider to be priorities for action.

Start date of proposal's development: 19 Dec 2022

Start date of CRWIA process: 13 May 2024

2. With reference given to the requirements of the UNCRC (Incorporation) (Scotland) Act 2024, which aspects of the proposal are relevant to children's rights?

By taking forward our actions to improve the lives of those living with a rare condition in Scotland, children and young people living with a rare condition, and their families, will benefit from a shorter diagnostic odyssey, healthcare professionals having greater awareness of rare conditions, improved co-ordination of care, and improved access to specialist care.

Our Action Plan and Progress Report will have a positive impact on children's rights, in particular Articles 2, 3, 6, 17, 23, and 24.

Please provide a summary of the evidence gathered which will be used to inform your decision-making and the content of the proposal.

Our Action Plan, and this Progress Report, were developed with input from Scotland's Rare Disease Implementation Board (RDIB), which includes representation from clinical geneticists, healthcare professionals and researchers and third sector organisations who support people with rare conditions.

Community engagement was facilitated by Genetic Alliance UK so that those living with a rare condition, and those involved in care and/or advocacy, would have their priorities at the centre of our Action Plan and in noting our priorities going forward with this Progress Report. This engagement included parents who cared for children with a rare condition. Genetic Alliance UK have also published several reports detailing the impact on families of caring for a child with a rare condition. Engagement was held to ensure that evidence from the Rare Diseases Framework matched the priorities of the community in Scotland, and to gather more detail on their lived experience. Feedback from this formed the development of our Action Plan and Progress Report.

Correspondence received from the rare community, including parents and carers, provided evidence on the experiences of those living with a rare condition and what the current issues are.

3. Further to the evidence described at '3' have you identified any 'gaps' in evidence which may prevent determination of impact? If yes, please provide an explanation of how they will be addressed.

We do not have, and did not seek as part of the development of the strategy or progress report, views of children and young people or data on their experiences of current services. However, community engagement did include parents and carers of children and young people with a rare condition, which we considered to have covered their views and needs to sufficient extent for our purposes.

Going forward we will continue to work with our third sector partners to seek out opportunities for children and young people, and their families to feedback and inform our rare conditions policy.

4. Analysis of Evidence

Our analysis of key reports showed:

- Genetic Alliance UK – [Good Diagnosis](#) report: outlines the importance of providing timely diagnosis so that patients and families can access effective care sooner.

The report details community feedback on themes including not being believed when reporting unfamiliar symptoms, or GPs not knowing what to do/what referrals to put in place, given how infrequently (if ever) they encounter a rare condition. Even if referral is given, multiple appointments are often required before diagnosis. Support after diagnosis, being well informed and knowing where to turn to were also highlighted as priorities.

- Genetic Alliance UK – [Stats Behind the stories](#) report: recommends that clear supportive information should be provided, and improvement work should encompass all rare conditions whilst taking the most common groups as examples in which good practice can be identified.

The Stats Behind the Stories report was discussed at our Rare Disease Implementation Board: the strategic grouping of conditions to provide common support was noted as aligning with our aim to identify good practice which could be applicable to a wide range of rare conditions by drawing out commonalities in the care required. This pragmatic approach continues to influence our priorities going forward. Identifying commonalities across groups of conditions also eases the task of designing care pathways, which we will bear in mind for future work in this direction.

- [UK Rare Disease Framework](#) and [Rare Disease Action Plan](#): set out four key priorities as identified by rare community and third sector: that people living with rare conditions benefit from faster diagnosis, from healthcare professionals having increased awareness, from better co-ordination of care and improved access to specialist care.

The UK Rare Diseases Framework was developed in extensive consultation with the rare community, and gathered extensive evidence on their lived experience of accessing diagnosis and effective care. The consensus that diagnosis could be delayed sometimes by years; that healthcare professionals lacked understanding to identify rare conditions; that appointments with specialists could be frequent and poorly co-ordinated, and that access to specialist care was limited, led to the four shared priorities that each UK nation was tasked with developing into an Action Plan.

Scotland's first Action Plan for Rare Diseases took the priorities of diagnosis, awareness, co-ordination and specialist care through a suite of engagement sessions, so that we could develop our Action Plan specifically from the identified needs of the rare community in Scotland. This was integrated into our Action Plan in the form of quoted feedback, followed by explanations as to how each of our actions would benefit people living with a rare condition.

Correspondence from parents of children with rare conditions demonstrated ongoing challenges with raising awareness of rare conditions and access to treatment and care.

Further engagement, again led by Genetic Alliance UK, took place in early 2024 to check that the community's priorities still aligned with our progress and our intended forward look. This informed the development of our Progress Report.

5. What changes (if any) have been made to the proposal as a result of this assessment?

Evidence from the most recent rare community engagement, and evidence prior to this, have informed our priorities for the year going forward. To maximise our resources, we are prioritising actions that will raise awareness among healthcare professionals of rare conditions. This is key to all community priorities, from reducing the time to diagnosis and time burden, ensuring people can make the most of appointments and feel listened to, to shortening the time to effective care. We are further progressing work to enable faster diagnosis by supporting the development of genomics services, and learning from work in the other UK nations to support the diagnosis of non-genetic conditions.

Conclusion

6. As a result of the evidence gathered and analysed against all UNCRC requirements, what is the potential overall impact of this proposal on children's rights?

Applicable answer(s): Positive

7. If you have identified a positive impact on children's rights, please describe below how the proposal will protect, respect, and fulfil children's rights in Scotland.

Children and young people living with a rare condition, and their families, will benefit from a shorter diagnostic odyssey, healthcare professionals having greater awareness of rare conditions, improved co-ordination of care, and improved access to specialist care.

Article 2: Non-discrimination: our work seeks to reduce healthcare inequalities that impact children with a disability and/or long term health condition.

Article 3: Best interests of the child: our work seeks to support better co-ordinated care provided by healthcare professionals who will have greater awareness of rare conditions through the resources that we create or signpost.

Article 6: Life, survival and development: our priorities are to enable faster diagnosis, increased awareness among healthcare professionals, better co-ordination of care and increased access to specialist care for everyone living with a rare condition. Rare conditions can be present at birth, and more prompt diagnosis and care can improve children's lives.

Article 17: Access to information: our work seeks to support more resources being available to people with a rare condition, their families and carers. Having a landing page on NHS Inform is the first instance of making public-facing information available about rare conditions.

Article 23: Children with a disability: our priorities are to enable faster diagnosis, increased awareness among healthcare professionals, better co-ordination of care and increased access to specialist care for everyone living with a rare condition. All of these enable higher quality care for children with a disability caused by a rare condition.

Article 24: Health and health services: our work aims to raise awareness of rare conditions among healthcare professionals, so that they can diagnose and treat rare conditions more promptly and effectively. Improved co-ordination of care and access to specialist care will also improve the standards of care for people with rare conditions.

8. If a negative impact has been identified please describe below. Is there a risk this could potentially amount to an incompatibility?

No negative impacts have been identified.

Mitigation Record

What options have been considered to modify the proposal in order to mitigate negative impact or potential incompatibility issues?

Please summarise mitigation actions taken below

Issue or risk Identified per article/ Optional Protocol

N/A

Action Taken/ To Be Taken

N/A

Date action to be taken or was taken

N/A

As a result of the evidence gathered and analysed against all wellbeing indicators, will the proposal contribute to the wellbeing of children and young people in Scotland?

Applicable answer: yes

If yes, please provide an explanation below:

Wellbeing Indicator: Healthy

Children and young people living with a rare condition, and their families, will benefit from a shorter diagnostic odyssey, healthcare professionals having greater awareness of rare conditions, improved co-ordination of care, and improved access to specialist care.

Faster diagnosis allows for earlier and more targeted intervention, lessening the disease burden borne by children with a rare condition and their families; third sector support and guidance can also be sought once a diagnosis is known. Greater co-ordination of care allows multidisciplinary teams to work together in ways that cut down on the time taken to travel to multiple appointments, and improved access to specialist care opens up a wider range of options for targeted intervention. Underpinning all of this is increased awareness of rare conditions among healthcare professionals, who are then better equipped to spot and diagnose rare conditions and able to refer children on to the correct care pathways.

9. How will you communicate to children and young people the impact that the proposal will have on their rights? (Guidance Section 3.2, page 27)

We will work with key stakeholders, including the Genetic Alliance UK, to ensure children and young people are informed about the Progress Report and impacts on them. The CRWIA will also be published on gov.scot alongside the progress report so will be in the public domain for those wishing to read it. In so far as possible the CRWIA has been written in accessible language so those reading it can understand its content and the potential impact identified in the assessment.

Post Assessment Review and sign-off

10. Planning for the review of impact on children's rights and wellbeing

We will continue to engage, through Genetic Alliance UK, with families affected by rare conditions to measure impact. CRWIA will be updated with subsequent progress reports or with new Action Plan/strategy, whichever comes sooner.

11. Sign off

Policy Lead Signature & Date of Sign Off: Alan Burns 6 August 2024

Deputy Director Signature & Date of Sign Off: Lynne Nicol 6 August 2024

Date CRWIA team first contacted: 06/08/2024



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