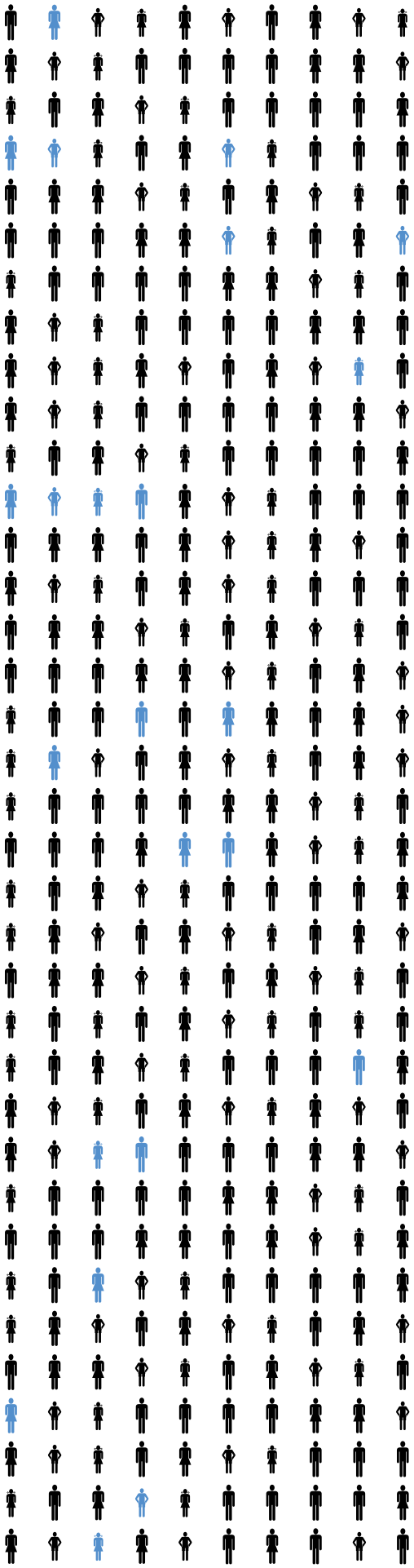
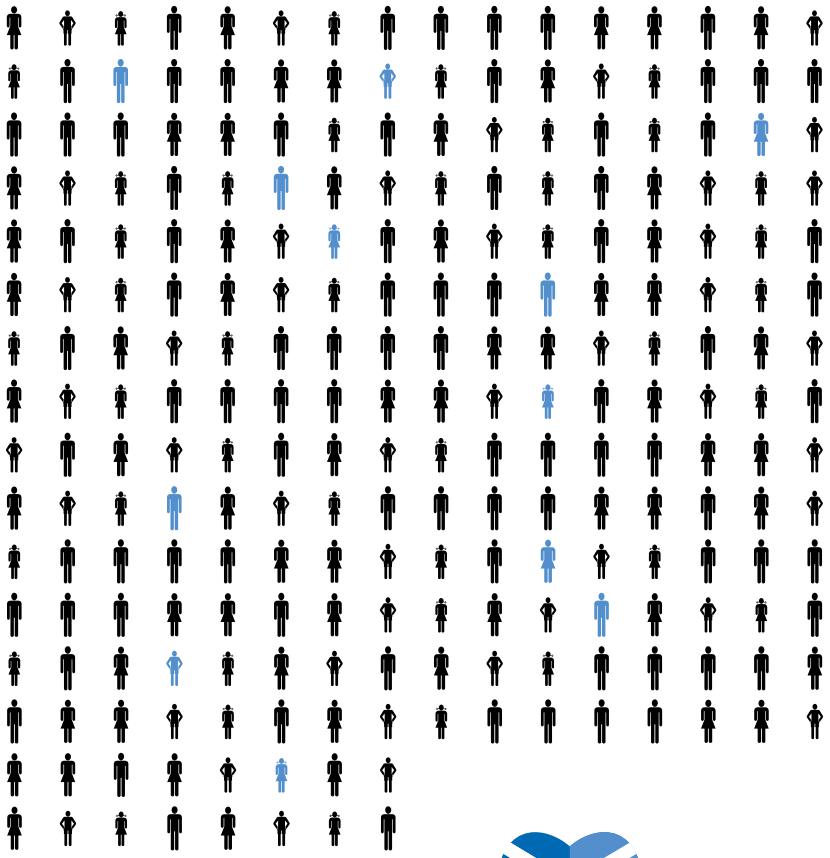
Rare Disease Scotland

Final Progress Report Summary

February 2021





Healthier Scotland

Report Summary

The Scottish Implementation Plan, It's Not Rare to Have a Rare Disease was first published in 2014 following the publication of the UK Strategy for Rare Diseases in November 2013. The title came from EU research statistics that showed whilst each rare disease only affects a small number of people (5 people or fewer in 10,000) there are actually between 6,000 – 8,000 rare diseases, and together these affect approximately 8% of the population. To put this in context, there are approximately 437,000 people out of a population of 5,463,300 in Scotland with a rare disease.

The Strategy included a list of 51 commitments and formed the blueprint for work across all the four UK nations to improve services, support patients and promote the role of research. Each nation developed their own implementation plan specific to the needs of their rare disease population.

The 51 commitments of the Strategy cover five specific themes:
- Empowering those affected by rare diseases
- Identifying and preventing rare diseases
- Diagnosis and early intervention
- Co-ordination of care
- The role of research.

Scotland published its own progress report against the 51 commitments in February 2018 which highlighted a variety of areas requiring further progress to be taken forward in order to meet the commitments by the end of 2020.

The final progress report evaluates the progress made against each of those 51 commitments originally set out in 2013 and reflects on the societal, economic and technological changes that have taken place since the publication of the original strategy.

The information in the report will inform our future rare disease policy to ensure any future strategy encompasses the advancements we are seeing daily in our healthcare system in Scotland.

Each chapter of the report highlights key areas of progress against the relevant commitments and a more specific breakdown of progress against can be found in the annex at the end of the report.

It should be noted that much of the progress made towards the 51 commitments has only been made possible by working in partnership with a range of organisations out with the Scottish Government. Working with those who understand and support the rare disease community most is a priority for the Scottish Government, ensuring the rare disease community is put at the heart of any policies that affect them.

There have been 3 key flagship deliverables. More detail on these can be found in the relevant chapters in the final progress report.

✓ **The Congenital Anomalies Register (CARDRISS)**

CARDRISS once fully established, will register babies affected by a major structural or chromosomal anomaly or recognised syndrome.

What does this mean for rare disease patients?

While the Congenital Anomalies Register is still being developed we have already seen a great benefit just by linking historical datasets to provide, for the first time in Scotland a record of congenital anomalies.

The dataset and the register are beginning to help inform the planning of services for individuals and families affected by congenital anomalies and rare diseases. Even more so once the Register is live, it will also allow NHS Scotland to support the prevention of anomalies where possible, understand the impact of antenatal screening and support research into these conditions.

✓ **The Bridge to a Scottish Strategy for Genomics**

Advancement of the strategy was driven by our partners in NHS Services Scotland National Services Division, the strategy supports the transition of genomic testing for inherited rare disease from the research setting into regular genetic testing services provided by the NHS in Scotland.

What does this mean for rare disease patients?

Scottish physicians and scientists have made world-leading contributions to research on rare disease genetics and genomics; and a well-established network of clinicians and clinical scientists have been delivering evidence-based genetic testing for NHS Scotland (NHSS) patients for over thirty years. Continuing advancements in genomics medicine improves a range of factors for rare disease patients included diagnosis, access to treatment and co-ordination of care.

✓ **The Rare Disease Forum**

The UK Wide Rare Disease Forum was hosted by Scotland in November 2019. The Forum is made up of a wide range of rare disease stakeholders including patients themselves, third sector organisations and clinicians.

What does this mean for rare disease patients?

This year's Forum heard presentations from a range of healthcare professionals, patient organisations and those affected by rare disease. The outcomes from these discussions, and breakout sessions and evidence gathered, will feed into the development of the next iteration of the Rare Disease Strategy post 2020 and more specifically Scotland's own Action Plan. The Forum highlights the need for ongoing collaboration between patients and policy makers in order to ensure policies truly reflect the needs of rare disease patients.

This publication is available at **www.gov.scot**

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