Minister for Public Health, Sport and Wellbeing
Joe FitzPatrick MSP

Scottish Government
Riaghaltas na h-Alba
gov.scot

T: 0300 244 4000
E: scottish.ministers@gov.scot

The Rare Disease Community

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Dear Rare Disease Community,

As we approach the thirteenth annual international Rare Disease Day on 29th February 2020 and recognise that we are now in the final year of the current UK Rare Disease Strategy, I would like to take an opportunity to update you on Scotland's position with regard to rare disease.

From the outset I'd like to assure the rare disease community that, post 2020, the Scottish Government remains absolutely committed to ensuring that all people living with a rare disease are able to access the best possible care and support, and benefit from healthcare services that are safe, effective and put the patient at the centre of their care.

With that in mind, I can confirm that Scotland will continue to work collaboratively with UK Government and other devolved nations on the development of a new UK wide rare disease Framework that will follow the UK Strategy for Rare Diseases when it expires at the end of 2020. Following this, we will then publish our own action plan setting out how we will implement the Framework for the rare disease community here in Scotland.

In July 2019, many of you will have taken part in a UK wide "National Conversation on Rare Disease". This took the form of a survey, which launched in October 2019 and ran for 6 weeks to gather the views from the rare disease community to help inform the new post 2020 framework. Over 6,200 responses were received from rare disease patients, healthcare professionals, researchers and industry.

I would like to thank all of our Scottish participants for making time to identify and share the major challenges faced by rare disease patients and the people and organisations that care for them. The themes identified in the survey will be integral to the development of the new framework mentioned above, and also Scotland's subsequent action plan.

While Scotland will publish a final progress report to co-incide with the conclusion of the strategy at the end of 2020, I would like to share a brief progress update on the 51 commitments.

Since the last published update in February 2018 the focus of the Rare Disease Strategic Oversight Group (RDSOG), with the support of a short life working group, has been on 3

clear priorities - co-ordination of care, raising awareness of rare disease amongst health professionals and research. I will set out some detailed progress against each of them.

Firstly, in relation to co-ordination of care, a short life working group has been established to explore the challenges and opportunities in delivering co-ordination of care for rare disease patients and to further develop the use case personas produced through a number of workshop collaborations. These are based on the real evidence of the challenges faced by rare disease patients, their families and healthcare professionals.

We continue with our work on raising awareness of rare diseases amongst healthcare professionals, particularly improving access to relevant information. Another short life working group is taking forward a range of actions including improving patient access to information through NHS Inform and the development of a rare disease tool kit for use by health care professionals. Furthermore, as part of the Scottish Government's Digital Health and Care Strategy, we are working with digital platform teams to enable improved access to information for health professionals.

In terms of research, we are establishing a Scottish Congenital Anomaly Register. The Congenital Anomalies and Rare Diseases Registration and Information Service for Scotland (CARDRISS) is part of a wider programme of work to improve information on individuals affected by rare diseases, and hence ultimately improve their outcomes.

The first report on congenital anomalies in Scotland was published on 26 November 2019. The information gathered from the register will allow a better understanding of serious conditions and inform the planning of services along with a range of further benefits. Work is ongoing and the register is expected to be ready to register babies with a congenital anomaly from 2021 onwards.

Scotland has also continued to engage with rare disease patients and the wider community through the third UK Rare Disease Forum Conference which we were delighted to host in Edinburgh on 29 October 2019. The conference provided attendees with an opportunity to hear from policy makers and leading clinicians on the of developments in rare disease research and genomic medicine. It also provided an opportunity for healthcare professionals, patients, patient organisations and government officials to discuss challenges and opportunities for improving rare disease patient care.

The Scottish Government has also invested in the advancement of genomics and precision medicine in Scotland. In 2018/19, we included a commitment in the Programme for Government, to support the continued development of genomic testing approaches in NHS Scotland by investing £4.2 million over the course of three years, building on the existing genetics capabilities of the NHS, and experiences gained through Scottish Genome Partnership (SGP) to evaluate different genomic testing approaches, facilitate continued collaboration with Genomics England, and improve genomic data analysis, sharing and storage within the NHS. An action plan is in place to evaluate the progress over the next two years to inform further strategic planning.

We have also established the Scottish Genomics Leadership Group in response to the first recommendation made by the published report from the Scottish Science Advisory Report (SSAC) in February 2019.

In an evolving landscape, I hope you will agree that real progress has been made in Scotland for the rare disease community. I'd like to assure you that, post 2020 the work will continue through the development of the UK wide Framework and our commitment to deliver

a new action plan which will benefit rare disease patients, their families and those involved in their care.

My officials will continue engaging with the rare disease community through a series of workshops and will undertake further evaluation of our progress against the existing 51 commitments throughout 2020. I hope you all take the opportunity to engage in that process.

The information and evidence gathered will provide a robust evidence base which will be considered by the Rare Disease Oversight Group.

I would expect that evidence base will shape our future rare disease action plan for Scotland.

I hope that this letter provides some comfort and reassurance of our ongoing commitment to the rare disease community.

Joe FitzPatrick MSP

Copied to:
Matt Hancock MP, Secretary of State for Health and Social Care, Department of Health, UK Government
Alister Jack MP, Secretary of State for Scotland
Vaughan Gething AC/AM, Minister for Health and Social Services, Welsh Government
Robert Swann, MLA, Department of Health, Northern Ireland