Cross-Party Group Annual Return

Name of Cross-Party Group
Cross-Party Group on Rare, Genetic and Undiagnosed Conditions
Date Group Established (the date of establishment is the date in this parliamentary session that the Group held its initial meeting, where the office bearers were elected and not the date that the Group was accorded recognition. All Groups should hold their AGMs on, or before, the anniversary of this date.)
16 June 2021
Date of Most Recent Annual General Meeting (AGM)
8 September 2022
Date Annual Return Submitted
22 September 2022
Date of Preceding AGM [this date is required to aid clerks in verifying that the most recent AGM has taken place within 11 to 13 months of the previous AGM]
16 June 2021
Group Meetings and Activities Please provide details of each meeting of the Group including the date of the meeting, a brief description of the main subjects discussed and the MSP and non-MSP attendance figures. Details of any other activities, such as visits undertaken by the Group or papers/report published by the Group should also be provided.

The Cross Party Group on Rare, Genetic and Undiagnosed Conditions has continued to play a vital role in bringing together key stakeholders from across Scotland to identify the main priorities for improving the experience of care for people with rare, genetic and undiagnosed conditions.

The CPG has proved to be an important forum for our underserved community, allowing small support organisations and people with rare conditions a valuable forum for discussion of the main challenges they face including, but not limited to: diagnosis, access to services, treatment and medicines, information and support, raising awareness with health professionals and coordination of care.

The CPG has utilised meetings in the 2021/22 period to inform the implementation of the UK Rare Disease Framework in Scotland and the development of the Scottish Government's Rare Disease Action Plan. Our meetings have allowed for expert presentations and discussions around the UK Rare Disease Frameworks four priorities and the discussions and recommendations have been shared with the Scottish Government's Rare Disease Team to inform their work. The CPG's report 'Improving Care for Rare Conditions in Scotland' has helped shape the direction of discussions of the Rare Disease Implementation Board and led to the creation of a Short Life Working Group on care coordination for rare conditions. The CPG has also played a pivotal role in advocating for better newborn screening for rare conditions, with a focus on Spinal Muscular Atrophy. This work has influenced the development of a research project in Scotland to roll out newborn screening for SMA.

It was noted that the CPG meetings had attracted large numbers of external attendees and whilst it had been a challenge to hold quorate meetings, the group had attracted non-member MSPs for some meetings.

The CPG will continue to inform the development of Scotland's Rare Disease Action Plan and in the 2022/23 session we will focus on:

- Improving the experience of diagnosis (including achieving faster diagnosis for rare conditions through genetic testing, newborn screening and raising awareness with health professionals)
- Improving the experience of care with a focus on care plans, coordinated care and workforce.
- Improving access to services, treatments and medicines for rare conditions for people living in Scotland.
- Improving access to high quality information and support for people with rare, genetic and undiagnosed conditions and the health and social care professionals that support them.

16 June 2021 – UK Rare Disease Framework

This meeting focused on the introduction of the UK Rare Diseases Framework and featured a presentation by the Scottish Government Rare Disease Team. The meeting also featured presentations from expert speakers including Professor Zosia Miedzybrodzka (diagnosis), Dr Martina Rodie (Care Coordination), Dr Catherine McWilliam (Newborn Screening).

It was noted by the Scottish Government Rare Disease Team that the work of the CPG would help to inform the development of the Scottish Rare Disease Action Plan.

MSPs in attendance: Bob Doris MSP, Paul McLennan MSP

External attendees: 23

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18 January 2022 – Improving Access to Specialist Care, Treatment and Medicines

This meeting focused on the challenges around accessing specialist care, treatment and medicines for rare conditions. The meeting featured powerful presentations from people living with Phenylketonuria and there was discussion about inequity in accessing treatment for the condition throughout the UK. Further examples were provided by Muscular Dystrophy UK and a broad conversation about the challenges of accessing medicines for rare conditions was had. The discussions fed into the Why Medicines Matter project, which produced a report that was shared with the Scottish Government's Rare Disease Team to inform the development of the Scottish Rare Disease Action Plan.

MSPs in attendance: 4 (Bob Doris MSP, Paul McLennan MSP, Monica Lennon MSP*, Neil Gray MSP**)

External attendees: 31

*Monica Lennon MSP is not a member of the CPG on Rare, Genetic and Undiagnosed Conditions.

**Neil Grey MSP was a member of the CPG on Rare, Genetic and Undiagnosed Conditions at the time of this meeting.

1 June 2022 – Newborn Screening in Scotland

The issue of newborn screening had been brought to the attention of the CPG by prominent clinicians and support organisations. To address uncertainty about the newborn screening decision making process in Scotland, Dr Tamsin Sommerfield (Consultant in Public Health Medicine for National Screening Programmes - National Services Division and National Clinical Advisor for Screening - National Screening Oversight) and Gareth Brown (Director of Screening, National Screening Oversight Clinical Directorate, NHS National Services Scotland) attended the meeting and provided a presentation on decision making in Scotland and the UK, including an update of an expected review of processes. Following the presentation, a wide ranging discussion was had and members of the rare, genetic and undiagnosed conditions community strongly voiced their support for increased newborn screening.

There was a specific discussion around newborn screening for Spinal Muscular Atrophy (SMA) and it was noted that following a letter written by the CPG to the Cabinet Secretary following the meeting in 2021, a research project was being developed to provide newborn screening for SMA in Scotland.

MSPs in attendance: 2 (Bob Doris MSP, Jackie Baillie MSP*)

External attendees: 27

*Jackie Baillie MSP attended in her capacity as Convener of the CPG on Muscular Dystrophy but is not a member of the CPG on Rare, Genetic and Undiagnosed Conditions.

22 June 2022 – AGM and Rare Disease Research

It has been intended that the AGM would be held during this meeting, but insufficient numbers of MSPs were in attendance.

This meeting featured a presentation by Professor Wendy Bickmore, Director of the Medical Research Council Human Genetics Unit, Institute of Genetics and Cancer at the University of Edinburgh described the work ongoing to understand rare diseases and how genetic variation impacts more common conditions. It was noted that this work would be vital to improving diagnosis of rare, genetic and undiagnosed conditions. It was agreed that a letter would be written to the Cabinet Secretary to ask what support was available to ensure this work could continue.

A presentation on the Why Medicines Matter project was given by Michelle Conway, highlighting how the views of the CPG had been taken into account during the development of the Why Medicines Matter report which had been shared with the Rare Disease Implementation Board.

MSPs in attendance: 1 (Bob Doris MSP)

External: 30

8 September 2022 - AGM

A meeting was held on the 8 September 2022, the earliest available opportunity, to undertake the AGM activities which could not be completed at the meeting on 22 June. Bob Doris MSP was agreed as Convener of the Group, Paul McLennan was agreed as Deputy Convener and Genetic Alliance UK will continue to provide Secretariat and Treasurer support.

It was noted that the CPG continued to provide a vital forum for the community affected by rare, genetic and undiagnosed conditions in Scotland and that the CPG has successfully informed the development of Scotland's Rare Disease Action Plan and had played a key role in supporting the development of the newborn screening research pilot for spinal muscular atrophy.

The challenges of holding meetings with a large number of MSPs was noted and it was agreed that discussion on how to increase membership would be undertaken alongside wider awareness raising work in the Scottish Parliament.

MSPs in attendance: 2 (Bob Doris MSP, Paul McLennan MSP)

External: 2

MSP Members of The Group

Please provide names and party designation of all MSP members of the Group.

- Bob Doris MSP
- Paul McLennan MSP
- Emma Harper MSP (as of 15 March 2022)
- Miles Briggs MSP
- Carol Mahon MSP
- Neil Grey MSP (until 25 January 2022)

Non-MSP Members of The Group

For organisational members please provide only the name of the organisation, it is not necessary to provide the name(s) of individuals who may represent the organisation at meetings of the Group.

Individuals	
Organisations	Genetic Alliance UK Alternating Hemiplegia of Childhood UK (AHC UK) Autoimflammatory UK Behcet's UK British Liver Trust CHAMP 1 EDS UK Haemophillia Scotland HSP Support Group MyAware NLRP12 Office for Rare Conditions (Glasgow) PNH Scotland PSP Association Rare Disease Nurse Network

	The Aarskog Foundation The Smith-Magenis Syndrome (SMS) Foundation Turner Syndrome Support Society

Group Office Bearers

Please provide names for all office bearers. The minimum requirement is that two of the office bearers are MSPs and one of these is Convener – beyond this it is a matter for the Group to decide upon the office bearers it wishes to have. It is permissible to have more than one individual elected to each office, for example, co-conveners or multiple deputy conveners.

Convener	Bob Doris MSP
Deputy Convener	Paul McLennan MSP
Secretary	Genetic Alliance UK
Treasurer	Genetic Alliance UK

If the Group is not disclosing any financial information please tick the box to confirm that the Group has considered the support received, but concluded it totalled under the threshold for disclosure (£500).

The CPG has not held an in person meeting during the 2021/2022 reporting period and therefore no costs for venues, catering, AV and printing have been incurred.

Genetic Alliance UK continue to provide Secretariat and administrative resource to the CPG. This equates to approximately 2 hours per week of Genetic Alliance UK's Policy and Engagement Manager time.

Subscription Charged by the Group

Please provide details of the amount charged and the purpose for which the subscription is intended to be used.

No subscription is charged.	
Convener Contact Details	
Name	Bob Doris MSP
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