GENETIC ALLIANCE UK

Report of the Board of Trustees and Accounts

2021-2022

GENETIC ALLIANCE UK

REPORT OF THE BOARD OF TRUSTEES

for the year ended 31 March 2022

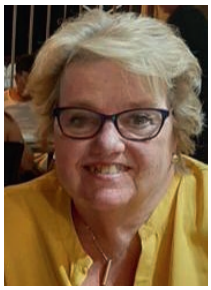
The trustees who are also directors of the charity for the purposes of the Companies Act 2006, present their report with the financial statements of the charity for the year ended 31 March 2022. The trustees have adopted the provisions of the Accounting and Reporting by Charities: Statement of Recommended Practice applicable to charities preparing their accounts in accordance with the Financial Reporting Standard applicable in the UK and Republic of Ireland (FRS 102) (effective second edition January 2019) and the Companies Act 2006.

CONTENTS

3 Welcome from the chair
3 Rupert's Story
4 Objectives and aims
5 About genetic, rare and undiagnosed conditions
6 **Making progress: The UK Rare Diseases framework**
7 **Building our community**
7 Genetic Alliance UK members
8 Supporting our members – Building Rare Resilience
10 SWAN UK
11 SWAN UK Cymru
13 **Building greater awareness**
13 Rare Disease Day 2022
16 Good Diagnosis Report
20 **Driving Progress**
22 Policy Engagement
23 Patient Empowerment Group (PEG)
24 Making our Members' Voices Heard
25 Wales
26 Rally for Rare
27 Scotland
28 Cross Party Group (CPG) on Rare, Genetic and Undiagnosed Conditions (Scotland)
29 Rare Resources
30 The Virtual Involvement Panel (Scotland)
31 Research
33 Building our organisation
34 The coming year
35 Treasurer's letter and financial review
36 Structure, governance and management
37 Reference and Administrative Detail
38 Trustee responsibility
39 Independent Auditor's Report to the Members of Genetic Alliance UK
42 Statement of financial activities including income and expenditure account
43 Balance sheet
44 Cash flow statement
45 Notes to the financial statements
60 Acknowledgements
61 Our Members

Genetic Alliance UK
contactus@geneticalliance.org.uk
geneticalliance.org.uk
Registered charity numbers: 1114195 and SC039299
Registered company number: 05772999

WELCOME FROM THE CHAIR



Following the publication of the UK Rare Diseases Framework at the start of the year, 2021/22 was a crucial year for Genetic Alliance UK and our community.

The Framework brings together many of our priorities including diagnosis, screening, and coordination of care. Throughout the year, we have engaged closely with the development of genomic medicine and the myriad of opportunities to improve access to medicines for rare conditions. We have put significant resources into making sure there are as many voices of people living with genetic, rare and undiagnosed conditions as possible feeding into the four nations' Framework implementation plans.

It is particularly important that I take this opportunity to thank our interim joint Chief Executives, Nick Meade, Director of Policy, and Lauren Roberts, Director of Engagement and Support for giving us the continuity and stability needed during and after the pandemic. Their commitment gave us the space and time needed to determine how to take forward the Chief Executive role for the longer term. Their unstinting efforts – and alongside them those of the rest of our amazing team – ensured that not only have we come through the most challenging of times, but we have yet again produced a range of reports and services for our members that can only be considered outstanding when viewed against the significance of the pandemic and the changed financial and fundraising background. It is thanks to all these efforts that we can look forward with renewed enthusiasm to working together on behalf of everyone affected by a genetic, rare or undiagnosed condition.

While this report is rightly and predominantly a retrospective of the past year it is also important that we look forward. At the time of writing we have recently been joined by our new Chief Executive, Louise Fish, who joins us from the Tuberous Sclerosis Association, just one of our many member organisations. Welcome, Louise.

While Louise's priority is to set Genetic Alliance UK's strategic direction and day to day focus for the next five years, our passion, as always, is to champion the voices of 3.5 million people living with genetic, rare and undiagnosed conditions in the UK.

The Board of Trustees and I very much look forward to working with all of you to drive our vision.

Elizabeth Porterfield
Chair
Genetic Alliance UK

Rupert's story

At just eight weeks old, our gorgeous boy Rupert was diagnosed with SMA Type 1B (Spinal Muscular Atrophy). SMA is a rare genetic neuromuscular condition that causes muscle weakness and wasting due to the loss of motor neurons.

Around 45 children are born with SMA Type 1 each year in the UK. Many doctors are not aware of the disease, which delays diagnosis. Every day is crucial, especially in Type 1 babies, where motor neurons die every minute.

Rupert was given a life expectancy of just seven months without treatment. He had deteriorated significantly in a matter of days, and there was a concern his liver was too weak for treatment.

Thankfully Rupert passed the criteria and became the second baby in London to be treated with Zolgensma, a gene therapy that replaces the defunct SMN1 gene. Zolgensma uses harmless, genetically engineered viruses to deliver functioning SMN1 genes, in order to increase SMN protein levels.

OBJECTIVES AND AIMS

Genetic Alliance UK champions the voices of people living with a genetic, rare or undiagnosed condition in the UK, their families, and the amazing charities and support groups that advocate for them. We are an alliance of over 200 patient organisations.

The objectives of the charity:

- Relieve persons affected by a genetic, and/or rare, and/or undiagnosed conditions;
- Advance the education of the public concerning genetic, and/or rare, and/or undiagnosed conditions in such ways as the trustees of the charity see fit.

We advocate for fast and accurate diagnosis, good quality care and access to the best treatments. We actively support progress in research and engage with decision makers and the public about the challenges faced by our community.

We run two long-standing projects:

- **Rare Disease UK**, a campaign focused on making sure the new UK Rare Diseases Framework is as successful as possible, and to ensure that people and families living with rare conditions have access to a final diagnosis, coordinated care and specialist care and treatment. Rare Disease UK is the official organiser of Rare Disease Day in the UK.
- **SWAN UK** (syndromes without a name), the only dedicated support network in the UK for families affected by a syndrome without a name – a genetic condition so rare it often remains undiagnosed.

Genetic Alliance UK's vision is a society in which people affected by genetic, rare and undiagnosed conditions receive excellent care and are empowered and supported in all aspects of their lives.

Our five-year strategy from 2018 to 2023 sets out a clear mission to work with organisations and individuals to ensure that the needs and preferences of all people affected by genetic, rare and undiagnosed conditions are recognised, understood and met. We have four strategic objectives:

- Driving progress
- Building our community
- Building greater awareness
- Building our organisation.

You can read our 2018 to 2023 strategy at www.geneticalliance.org.uk/our-strategy/

We will begin reviewing this strategy in Autumn 2022, ready to approve a new five-year strategy in Autumn 2023 that will inform our business planning for 2024 and into the future.

Public benefit

The Board of Trustees confirm they have had regard to the Charity Commission's guidance on public benefit and have complied with their duty under section 4 of the Charities Act 2011 when reviewing the charity's aims and objectives; and in implementing current and planning future activities.

SWAN UK
syndromes without a name

RARE DISEASE UK

ABOUT GENETIC, RARE AND UNDIAGNOSED CONDITIONS

While genetic, rare and undiagnosed conditions are individually rare, taken together there are around 3.5 million people living with these conditions in the UK. This compares with 900,000 people who live with Alzheimer's disease in the UK and around 375,000 new cancer cases in the UK every year.

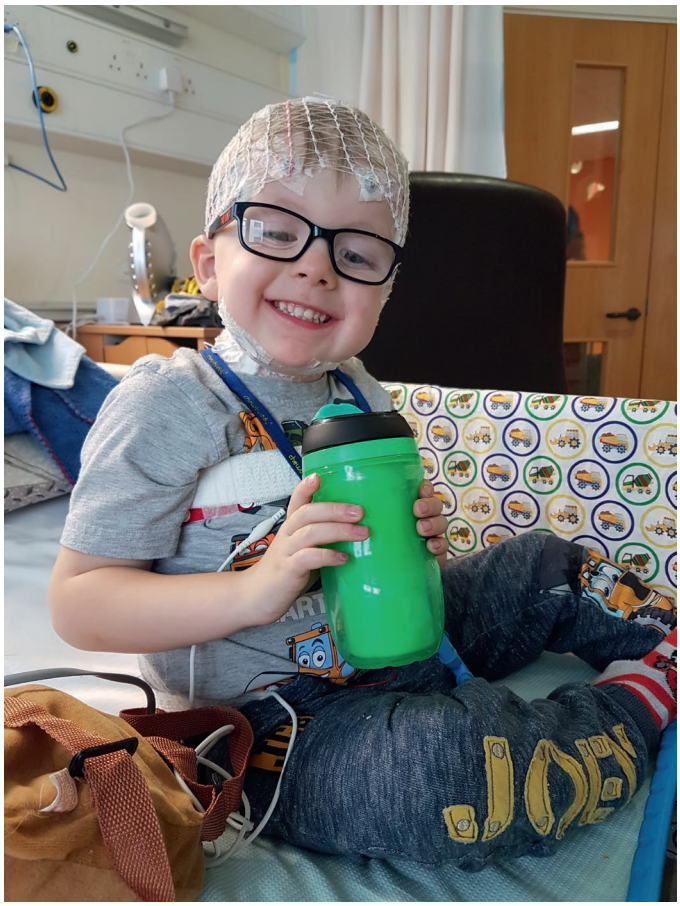
A **genetic condition** is one that is caused by a change in an individual's genome. The most common genetic conditions such as sickle cell disease affect around 15,000 people in the UK, with 300 babies born with the condition each year. The rarest genetic conditions may affect just one family in the UK, and there may only be a handful of people affected across the globe.

A **rare condition** affects fewer than 1 in 2,000 people. There are over 6,000 known rare conditions which can be life threatening and take longer to diagnose and treat than more common conditions. Eight out of 10 rare diseases have a genetic origin.

An **undiagnosed condition** is known as a 'syndrome without a name' or SWAN. Each year around 6,000 children are born in the UK with a genetic condition so rare that it does not yet have a name. This might be because the right test has not been developed to diagnose it, or the genetic cause of the condition has not yet been discovered.

Genetic, rare and undiagnosed conditions are often life-limiting and life-threatening. Seven out of 10 rare diseases affect children, and sadly more than three out of 10 children with a rare disease die before their fifth birthday.

People living with genetic, rare and undiagnosed conditions and their families can face a lifetime of complex care. They need vital support from the NHS, social care and education services to live their lives to the full.



MAKING PROGRESS: THE UK RARE DISEASES FRAMEWORK

Despite Covid-19 continuing to take up a large amount of bandwidth in national policy priorities and the health service budget, we were delighted to enter this year with a new UK Rare Diseases Framework published in early 2021 by the UK governments. You can find the UK Rare Diseases Framework at www.gov.uk/government/publications/uk-rare-diseases-framework.

The 2021 UK Rare Disease Framework set out the commitment of UK governments to improve the lives of people with rare conditions over the next five years. The Framework focuses on four priorities identified in Genetic Alliance UK's Rare Experience 2020 report, which are faster diagnosis, increased awareness of rare diseases, better coordination of care, and improving access to specialist care, treatments and drugs.

Genetic Alliance UK's report paints a detailed and vivid picture of a wide range and variety of experiences of people living with rare conditions in the UK. It was structured to match our expected themes of the incoming UK Framework for rare conditions, and included recommendations and learning points for the future of rare disease policy in the UK.

The publication of the Framework was the product of hard work by supporters of our Rare Disease UK campaign and thanks to the commitment of the whole multi-stakeholder rare conditions community. Our key aim this year has been to build on this opportunity, ensuring the voice of people living with rare conditions is heard loud and clear in the development of each individual Rare Disease Framework action plans for England, Scotland, Wales and Northern Ireland.

Both England and Northern Ireland's action plans were published by the end of March 2022. Well-informed and vocal members of the rare condition community are sitting on the relevant committees and working groups. Our work to support the development of a strong and deliverable action plan for each nation is set out throughout this report.

BUILDING OUR COMMUNITY

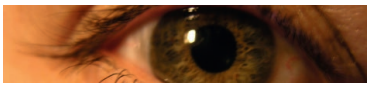
GENETIC ALLIANCE UK MEMBERS

Genetic Alliance UK drives improvement in diagnosis, treatment and care by speaking with a strong, united voice on behalf of people living with rare, genetic and undiagnosed conditions, their families, and the charities and support groups that advocate for them.

Genetic Alliance UK was delighted to welcome 15 new members in 2021/22. Membership of Genetic Alliance UK is open to patient organisations and not-for-profit organisations associated with genetic conditions. This enables us to represent a wide range of people affected by genetic conditions and campaign for the broad issues which affect all of our members.

A full list of members can be found on page 61.

The Aortic Dissection Charitable Trust



Action on Gilbert's Syndrome

CSNK2A1 Foundation

Glut1 Deficiency UK

Great Ormond Street Children's Charity

Acrodysostosis Support & Research

FAST UK – Foundation for Angelman Syndrome Therapeutics

SCN8A UK & Ireland

ITP Support Association

WomenZone

Fop Friends (Fibrodysplasia Ossificans Progressiva)

CASK Research Foundation

Thrombotic Thrombocytopenic Purpura Network (TTP Network)

CureGRIN Foundation

SMC1A Foundation

SUPPORTING OUR MEMBERS – BUILDING RARE RESILIENCE

4 courses | 32 participants | 25 member organisations

'This was a life changing experience for me for the positive and just what I needed.'

Over the year we collaborated with RareMinds to run four courses of our Building Rare Resilience programme. The programme affords staff and volunteers at Genetic Alliance UK member organisations the opportunity to take part in 12 weekly sessions of group counselling. The sessions are run by a counsellor specialising in providing professional counselling and wellbeing resources to rare organisations' staff and volunteers and aim to enable the individuals participating to build their emotional resilience and wellbeing.

Overall 32 individuals from 25 different member organisations attended across the 4 groups. All four groups have continued to meet regularly after their 12 weeks of official sessions ended, which is testament to the success of the programme.

Taking part in Building Rare Resilience – Phillippa's Story

Duchenne Family Support Group (DFSG) is a small charity (celebrating 35 years in 2022) that supports families who have children with the condition known as Duchenne Muscular Dystrophy. This is a rare genetic condition that mainly affects boys (as it is carried on their X chromosome) and is life-limiting. The muscles lack the protein known as dystrophin and so over time the boys' muscles waste away. Usually by the age of seven to nine they will need a manual wheelchair, by mid-teens they will need an electric wheelchair and as they get older, they will need cardiac checks, help with all forms of personal care, including toileting and some may need a catheter fitted, ventilation and respiratory support. They may also need help with feeding and will need a PEG fitted.

In November 2019, just before the pandemic started, Phillippa lost her son Daniel who lived with Duchenne Muscular Dystrophy at age 27. Phillippa works as a Development Officer at DFSG so she can help other families affected by the condition, and she decided to carry on even though it was a struggle as she coped with her grief. Although she misses Daniel dreadfully, he was always proud of the work that DFSG and Phillippa did to speak out on behalf of others.

When the Building Rare Resilience course was offered Phillippa decided to take part as she was not only struggling personally with her mental health, but she was also figuring out how to carry on working from home supporting the DFSG community and yet still be effective. She was exhausted from the continual Zoom meetings and social media messages, but since many families were struggling with their own journeys, she was hoping to talk to others in similar situations and to receive and give support going through the pandemic and beyond.

Phillippa felt the first meeting was a challenge, however, as soon as everyone got to know each other with the help of the facilitator and began sharing the things that were bothering them, everyone soon began to make new goals, relook their working practices. They were supported to continue to do their jobs, but much more effectively. As a result of these meetings, Phillippa's small group became really connected to each other that even when the course finished, they continued to meet weekly and support each other.

'We have learnt so much from each other, shared top tips, helped each other when the going got tough, advised courses of action where necessary and been like a family to each other. We were extremely lucky to have built such a close network and we still to this day continue to meet weekly. I for one would hate this to stop'. *Phillippa

Community Check-ins

38 sessions held | 244 attendees | 140 member organisations represented

Our weekly virtual Community check-ins are the bedrock of our engagement work with our members allowing us to keep in regular dialogue about issues arising affecting our community. 38 sessions were held over the year (breaks were taken over the summer, Easter and Christmas holiday periods). Overall 244 different individuals attended the sessions, representing nearly 140 Genetic Alliance UK Member organisations.

Check-in topics included:

Good Diagnosis report
vimeo.com/689597864

England action plan for rare diseases
vimeo.com/687173960

Whole Genome Sequencing in the newborn (Genomics England)
vimeo.com/639586563

Barriers and facilitators for a rare disease diagnosis
vimeo.com/619040458

NHS Medicines Repurposing Programme

Running Awareness days

Covid-19 vaccines and vulnerable children (RCPCH)
vimeo.com/577207048

Genomics and the UK Rare Diseases framework

Succeding in the digital world as a small charity
vimeo.com/694874196

SWAN UK

SWAN UK (syndromes without a name) is the only specialist support network available in the UK for families of children and young adults affected by a syndrome without a name. We work with families of affected children and young adults aged 0–25, providing support and information in hospital, at home and in their local communities.

3.7k members | 2.6k families | 6 virtual events | Dads Summit | Parent Rep workshop

Over the reporting period we have welcomed a number of new SWAN UK members, bringing the total membership figure to 3,686.

The decision was made to run a hybrid programme of events for SWAN UK members with the continuation of online events to cater for those families who continued to shield their vulnerable children from Covid-19 as well as some face-to-face events for families who felt ready to meet up. Over the course of the year, six virtual events were held, including a SWAN UK Virtual Information Event that had over 100 people sign up to attend. Physical events were reintroduced in September once changes in Covid-19 restrictions permitted it and three physical events were held from September to the end of the reporting period. These events included a SWAN UK Dads Summit which was attended by 13 members and our SWAN UK Parent Rep Workshop which was attended by eight Parent Reps.

A SWAN UK Couples Counselling programme was launched in September in collaboration with RareMinds. Over 40 couples registered their interest in taking part, and during the reporting period 15 couples were able to take part on a first-come-first-served basis.

Additionally, 10 virtual Zoom catch-up sessions were run for SWAN UK dads and 11 rounds of virtual coffee meets were arranged for members.

During the reporting period we've started to rebuild our outreach programme, focussing on raising awareness of SWAN UK among other professionals working in the health and social care sectors. SWAN UK presentations have been given to a number of organisations, including:

- Institute of Health Visiting
- Clinical Genetics, University Hospitals Bristol and Weston NHS Foundation Trust
- Neurodisability Team, South Tyneside and Sunderland NHS Foundation Trust
- Lincoln Parent Carer Forum

Undiagnosed Children's Day

Our awareness day, Undiagnosed Children's Day, took place on Friday 29 April 2022. As always this was an opportunity to share memories and experiences for our SWAN UK families. This year's day was special as we also celebrated SWAN UK's 10th birthday on 9 May.

SWAN UK CYMRU

139 members | 98 families | 18 parents attended info event | 12 participants at pamper event

SWAN UK membership in Wales continued to increase throughout the reporting period and in the first year of the project membership numbers in Wales rose at nearly double the rate of the rest of the UK. Our previous post-holder left the position of Engagement and Support Officer (SWAN UK – Cymru) at the end of September and after a successful round of recruitment Amanda Thomas started in the role at the beginning of January 2022.

Several successful online events were held including a UK wide virtual information day in April with a Wales specific breakout room. The event was well attended with 18 Welsh families and speakers attending the information session. In June a SWAN UK Cymru mum's pamper event was held online with 12 sign ups. Five mums attended live and seven mums participated by watching the event recording. Since this, site visits have been made in preparation for the role out of a physical family event.

The project's outreach has quickly gained momentum over the year and relationships have been forged with several key partners such as Contact Cymru, Tŷ Hafan Children's Hospice and Same But Different. The SWAN UK Cymru project was also represented at meetings for the new Welsh Undiagnosed Research Hub and continued to be involved with the newly-launched SWAN Clinic, including feeding into the development of patient evaluation material. A presentation on SWAN UK Cymru was given to paediatricians in South Wales as part of a Paediatric Study Day that was held in January. SWAN UK – Cymru has also been represented at the Wales CPG, Rare Disease Day Reception at the Senedd and Wales Gene Park's Genomics Cafe. These have all proved fruitful for networking opportunities, enabling the dissemination of information about the project through word of mouth.

Rare Resources Cymru (an early intervention information toolkit for families affected by rare or undiagnosed conditions) were translated into Welsh and designed ready for a year two roadshow.


WAN

BUILDING GREATER AWARENESS

RARE DISEASE DAY 2022

451.3k Twitter impressions | 12.3k Facebook reach | Website page sessions up 98% from Jan 2022

'I just wanted to say a big thank you to all of you for not only providing an amazing platform to amplify our voice on Rare Disease Day but for supporting us and rare diseases generally in everything you do.'
Rare Disease Day social media takeover participant

Rare Disease Day 2022 took place on Monday 28 February. It was celebrated in over 85 countries worldwide to raise awareness and generate change for 300 million people living with a rare condition, and their families and carers. Despite significant world events, #RareDiseaseDay2022 was trending on Twitter for most of the afternoon and evening. Engagement was high across all our platforms and participants in our social media take over slots reported similar increases in traffic to their channels.

'Our Twitter engagement increased threefold (by 300%) on our Rare Disease Day Takeover Day.'
Social media takeover participant

Rare Disease Day videos

4 videos | Over 5k views

Covid-19 has been extremely challenging for the UK's rare community with shattered support routines, reduced access to treatment and healthcare, and the increased isolation of shielding. The pandemic also shone a light on the increased health inequalities experienced by many marginalised groups in the UK, especially those from a black or minority ethnic community. Young people had their lives severely disrupted by schools and colleges closing. For many in the rare community, their rare condition is just one of many factors they are having to manage.

This Rare Disease Day we wanted to shine a light on these often unheard stories, while also demonstrating solidarity across the UK, Europe and Internationally.

Rare repository

Rare repository launched | Over 100 stories shared

Over 100 people shared their story on our new rare repository to light up a virtual window for Rare Disease Day. You can read their stories by visiting www.raredisease.org.uk/rdd2022/rare-repository/.

Shining a light on rare conditions

This year we partnered with ITN Productions Industry News to produce 'Shining a Light on Rare Conditions', a news-style programme raising awareness of people affected by rare, genetic and undiagnosed conditions. You can see the news programme at geneticalliance.org.uk/gauk-news/news/shining-a-light-on-rare-conditions/.

Light up for rare

Private homes, public buildings and landmarks including the Department for Health and Social Care, the Kelpies and the Gateshead Millennium Bridge were lit up in the Rare Disease Day colours to express support for the day.

Rare Disease Day parliamentary events

3 events | 150 participants |
Hosted by Russell George MS | Liz Twist MP |
Bob Doris MSP

'Thank you all, it's been a great session and to the amazing speakers.'
Esther Florence, The Pituitary Foundation

Unlike in 2021, individual virtual receptions were hosted for the Welsh, and the Scottish parliament and Westminster so that discussion could focus on issues for rare conditions relating to each nation. The receptions focussed on the theme of diagnosis, hearing from a variety of speakers on their experience of the diagnostic odyssey for rare conditions. The events were attended by around 200 people in total.

The receptions had an array of speakers including people with rare conditions. We were also delighted to receive addresses from the respective health ministers: Maria Caulfied MP, Eluned Morgan MS and Maree Todd MSP who looked forward to the release of the rare disease action plan throughout the following year. Attendees appreciated the opportunity to network in a virtual capacity and talk to the speakers after they had presented.

Genetic Alliance UK presented the Good Diagnosis report on page 16 at each of the receptions which was well received by the rare condition community. The report collated and materialised the experiences around diagnosis for people with rare conditions in a format that could be shared with policy makers. There was a strong interest in the recommendation for a Good Diagnosis Rare Conditions Patient Charter.

GOOD DIAGNOSIS REPORT

The lack of awareness about genetic, rare and undiagnosed conditions generally leads to slower referral, slower diagnosis, misdiagnoses and slower access to appropriate specialised care. Too often people living with the condition and their desperate families are left to research their own symptoms, or diagnosis, and this can lead to people finding unreliable, potentially alarming information which they have to digest without the support of a healthcare professional who understands the condition.

When an individual has a diagnosis of a rare condition that their principal healthcare professional does not know a lot about, they may continue to experience delays in being started on the correct care or treatment pathway, and they may experience delays in accessing appropriate services, medicines or research.

To coincide with Rare Disease Day 2022, Genetic Alliance UK launched, 'Good Diagnosis: Improving the experiences of diagnosis for people living with rare conditions'. Building on the findings of our Rare Resources (Scotland) and Rare Experience 2020 projects, we asked people to reflect on their experiences at three key stages of their diagnosis journey: their search for a diagnosis, receiving their diagnosis, and following diagnosis. The report identified the key principles of good diagnosis: a good diagnosis is timely and accurate, informed and supported, coordinated and collaborative, acknowledged and respected.

In the coming year we will work to implement the recommendations of the report:

- Increase awareness of rare conditions among healthcare professionals
- Ensure people with rare conditions have access to appropriate information and support throughout their diagnosis journey
- Develop a good diagnosis patient rights charter

Having to fight for the right care or treatment can be extremely challenging, particularly when unwell. The fight can be harder when a person is unsure about what rights they have, or who can support them. It is vital that people living with rare conditions and their families are aware of what to expect from the diagnosis journey, aware of their rights and how they can challenge decisions.

The Good Diagnosis report recommends that a Rare Conditions Good Diagnosis Patient Rights Charter should be developed to clearly communicate the standard of care people with rare conditions should expect to receive. We will be working closely with individuals, families and member organisations to listen to their views on what it should look like.

You can read the Good Diagnosis Report at geneticalliance.org.uk/gauk-news/news/good-diagnosis

GOOD DIAGNOSIS INFOGRAPHIC

Communications

Twitter reach 254k | Facebook reach 153.5k | 1,200 new Instagram followers

We have made significant strides in utilising our different platforms in an increasingly strategic way, curating content so that we are making the most of what each platform has to offer and engaging different audiences.

Over the last year we have maintained a strong presence on social media and had a high level of engagement across our channels. To illustrate this, the combined profile visits for all of our Twitter accounts was 254,381. Our combined Facebook page reach for all the channels was 153,515 and new followers across our Instagram accounts was 1,200. In addition, at the end of March 2022 we had 1,596 LinkedIn followers.

Social media takeovers

We regularly arrange for our social media channels to be 'taken over' by people with rare conditions, their families and the member organisations that support them. This helps our smaller member organisations and individuals living with the rarest conditions to increase their social media reach and gain new followers by communicating directly with the people who are following Genetic Alliance UK, Rare Disease UK and SWAN UK.

'Thanks again for this wonderful opportunity to raise awareness. We are so grateful to have your support ....' Marfan Trust

In addition to Rare Disease Day and Undiagnosed Children's Day, we have been working closely with our members to highlight the work that they do to support people with genetic, rare and undiagnosed conditions.

'Thank you so much for having us, I think it was a great thing to do to raise awareness of Jnetics and what we do …'

Website

882.4K website views | 20 Genetic Alliance UK member articles | 15 rare stories

Our website traffic has increased significantly demonstrating our high-quality content and promotion through our social media channels. Our websites received a combined 882,421 page views. We have collaborated closely with members and published more than 20 articles raising awareness of the conditions that they support as well as publishing 15 stories on Rare Disease UK highlighting the impact that rare conditions have on people and their families.


MANTIS

DRIVING PROGRESS

ALL PARTY PARLIAMENTARY GROUP ON RARE, GENETIC AND UNDIAGNOSED CONDITIONS (ENGLAND)

Genetic Alliance UK provides the secretariat to the APPG for Rare, Genetic and Undiagnosed conditions, which is chaired by Liz Twist MP. Thanks to Liz's excellent chairing and commitment this year, we brought some key issues to the attention of parliamentarians.

APPG: Access to medicines (England)

April 2021 | 14 parliamentarians | 4 speakers | NICE and NHS England attendance

'I thought that was a very good meeting today with signs that progress is being made'

14 parliamentarians including the Minister in charge of rare diseases, Lord Bethell, joined the meeting.

Guest speakers included: Kye Gbangbola (Sickle Cell Society), Kate Learoyd (National Society for Phenylketonuria), Emily Crossley (Duchenne UK), Jess Hobart (The UK Mastocytosis Support Group) and Roanna Maharaj (UK Thalassemia Society).

Speakers highlighted the many challenges they faced in accessing rare disease medicines.

Also in attendance was Meindert Boysen, Deputy Chief Executive and Director of the Centre for Health Technology Evaluation at NICE and Nina Pinwill, Head of Commercial Operations, Commercial Medicines Directorate at NHS England and NHS Improvement.

Discussions considered health inequality and the need for improvements in educating healthcare professionals. Attendees questioned when decision making bodies, such as NICE committees, would become more representative of the population and who should be responsible for ensuring data is collected in a way that is suitable for decision makers as the burden is often on people affected by rare conditions. Watch the meeting vimeo.com/542729454

'Thank you for inviting me to the Rare Disease APPG. It was interesting and very valuable.'
Head of Office for Mr Andrew Lewer MBE MP

Newborn screening (England)

June 2021 | 8 parliamentarians | 7 speakers | 3 key policy asks agreed

In June 2021, the APPG held a meeting on newborn screening. Eight parliamentarians and their representatives were in attendance to hear from a number of guest speakers, including: Professor Simon Heales (Great Ormond Street Hospital), Professor Laurent Servais (UK Newborn Screening Alliance & Muscular Dystrophy UK Oxford Neuromuscular Centre), Dr Will Evans (Niemann-Pick UK), Amanda Mortensen (Batten Disease Family Association), Karen Harrison (Alex – The Leukodystrophy Charity), Mandy Sanderson (Max Appeal) and Georgina Morton (ArchAngel MLD Trust).

Attendees agreed on the need for:

1. **The formation of a dedicated team of newborn screening experts to solely evaluate conditions to be added to the newborn screening programme.**
2. **The establishment of a streamlined evidence review process for evaluation of conditions to be added to the UK newborn screening programme.**
3. **A timeline to ensure appropriate efficiency and accountability for this process.**

Attendees also called for regular, meaningful and transparent engagement with stakeholders. Watch the meeting https://vimeo.com/561340806.

APPG: Parliamentary Outreach (England)

October 2021: 6 parliamentarians | 2 speakers | Update meeting – NICE Methods Review | newborn screening landscape | UK Rare Disease Framework

The APPG held a short meeting in October 2021 to provide an update on the NICE Methods Review, the newborn screening landscape and the UK Rare Diseases Framework. Six MPs and their representatives attended the meeting to hear from guest speakers including: Jess Hobart (The UK Mastocytosis Support Group (Masto UK)) and Karen Harrison (Alex – The Leukodystrophy Charity).

The MPs in attendance asked for more information on the differences across the nations in relation to the UK Rare Diseases Framework.

For this meeting, we launched the DoGooder tool which can be used by people in the rare community to contact their local MP, asking them to express interest in the APPG and broader activities related to rare conditions. You can see the DoGooder tool in action at geneticalliance.org.uk/gauk-news/news/rareappg-get-mps-to-support-people-living-with-rare-genetic-and-undiagnosed-conditions/. There are now also tools that can be used in the Welsh, Scottish and Northern Irish parliaments which altogether have been used over 200 times. This tool has helped the cross party groups to reach a wider group of members of parliaments and engage the rare community in our parliamentary activity.

APPG: The UK Rare Diseases Framework (England)

December 2021 | 7 parliamentarians | 3 speakers | hosted in conjunction with APPG for life sciences

In December 2021 the APPG held a meeting in conjunction with the APPG for Life Sciences, looking into the UK Rare Diseases Framework. Seven MPs, Lords and their representatives joined to hear from a number of speakers from both APPGs including: Kerry Leeson Beavers, (Alstrom Syndrome UK), Victoria Barrett, (The Association of the British Pharmaceutical Industry); and Jon Neal, (Takeda).

The attendees heard from a variety of stakeholders that sat on the bodies that were working on the development of the England Action Plan.

POLICY ENGAGEMENT

4 'Policy, Policy, Policy' community check-ins | 2 consultation webinars

Community check-ins

- 4 x 'Policy, Policy, Policy!' sessions hosted.
- Public affairs activities and the Rally for Rare campaign
- Our work with the Human Fertilisation and Embryology Authority (HFEA) on preimplantation genetic testing for monogenic disorders (PGT-M)
- Patient and public involvement in decision making
- UK Rare Diseases Framework and nation specific action plans
- Relevant policy consultations and other opportunities for involvement

Consultation webinars

- Innovative Medicines Fund (IMF) consultation
  'Thank you … very informative and objective, but most of all helpful.'
  IMF consultation webinar participant
- Webinar hosted: NICE Methods and Processes review

PGT-M statements for Human Fertilisation and Embryology Authority

50 statements submitted | 46 licences approved

HFEA feedback: Genetic Alliance UK statements made the condition 'come alive' and had a 'direct impact' on the committee's decision.

50 statements on preimplantation genetic testing for monogenic disorders (PGT-M) were provided to the Human Fertilisation and Embryology Authority (HFEA). Genetic Alliance UK reached out to the community to put together statements that portray the patient experience of living with a specific genetic condition. These statements are used as evidence when a committee at the HFEA makes the decision on whether to grant a PGT-M license for this condition, enabling families to have the choice of having a biological child without passing on the genetic condition.

PATIENT EMPOWERMENT GROUP (PEG)

14 members | 9 meetings |
Key recommendations report produced for UK Rare Diseases Framework

The Patient Empowerment Group (PEG) is a collaborative forum which enables patient representatives with an interest in rare and genetic conditions to come together to support one another in their efforts to represent people living with rare, genetic and undiagnosed conditions. PEG members from across the UK play a key role in advising on the development of the action plans which will implement the UK Rare Diseases Framework

Nine meetings were held during 2021/22, five of which reviewed the four priorities and the underpinning themes outlined in the UK Rare Diseases Framework. The meetings highlighted activity already happening and identified specific areas of interest and concern for the rare condition community under each of the priorities. A report was produced to outline key recommendations taken from these meetings which were fed into the delivery groups that were developing each nation's action plan.

Subsequent meetings focussed on scrutinising the implementation of the UK Rare Diseases Framework and ensuring that the voice of the rare condition community was effectively represented in the action plans. Members sitting on the delivery groups brought points of interest to the PEG meetings and received opinions from the rare condition community so that they could feed in a more representative voice to the development of the action plan. It also provided a space for cross-border collaboration.

The PEG members also acted as a sounding board for policy work at Genetic Alliance UK, contributing their insight on a wide variety of issues including the Down Syndrome Act, the Good Diagnosis report, the Rare Disease Day parliamentary receptions, and best practice when recruiting to roles for people with experience of living with rare conditions.

MAKING OUR MEMBERS' VOICES HEARD

Genetic Alliance UK staff sit on numerous committees, boards and steering groups to represent and champion the needs of the UK's genetic, rare and undiagnosed community.

- Breaking Down Barriers Advisory Board
- British Society for Gene and Cell Therapy Stakeholder Panel meeting
- British Society for Genetic Medicine Consensus Guidance development group for Pre-implantation Genetic Diagnosis (PGD)/ Pre-natal Diagnosis and (PND) for Cancer Susceptibility Genes
- Charity Medicines and Access Coalition
- Congenital Anomalies and Rare Diseases Registration and Information Service for Scotland Expert Advisory Panel (Scotland)
- Decipher Project Data Access Committee
- Delivering genomic sequencing in clinical practice: a patient-centred evaluation of the new NHS Genomic Medicine Service – Project Advisory Team
- Department of Health and Social Care UK and Sweden roundtable
- EURORDIS Council of National Alliances
- EURORDIS Membership Meeting 2021
- EXPRESS Study PPI group
- Genetic Counsellor Registration Board
- Genetics Laboratory Management Review Group (Scotland)
- Genomics England Newborns Ethics Working Group
- Genomics England Public dialogue on whole genome sequencing for newborn screening – oversight group
- Genomics Leadership Group (Scotland)
- Genomics Partnership Wales Programme Board
- Manchester University MSc in Advanced Therapy Medicinal Products
- National Genomics Board
- National Institute Health Research Advanced Therapy Medicinal Product Coordinating Group
- National Patient, Public and Professional Involvement Group (Scotland)
- Newborn Screening Collaborative
- NHS England – Accelerated Access Collaborative Advanced Therapy Medicinal Product Patient and Public Involvement and Engagement Working Group
- NHS England – Genomics People and Communities Forum
- NHS England – Genomics Programme Board
- NHS England – Patient and Public Voice Assurance Group
- NHS England – Preimplantation Genetic Diagnosis Commissioning Policy Development Group
- NHS England – Specialised Commissioning Stakeholder Forum
- NICE Guideline Committee – Children and young people with disabilities and severe complex needs
- NICE Methods review – working group and task and finish groups
- Office for Rare Conditions (Glasgow) Steering Group
- Preimplantation Genetic Testing Expert Panel (Scotland)
- Public Involvement Network Advisory Group Meeting (Scottish Medicines Consortium)
- Public Policy Projects Rare Disease Coalition
- Responding to genetic risk associated with customary consanguineous marriage
- Scotland Rare Disease Implementation Board
- Screen4Care Patient Advisory Board
- Solve RD Community Engagement Task Force – Steering Group Meeting
- UK Rare Disease Framework Board
- UK Rare Disease Stakeholder Forum
- Wales Rare Disease Implementation Group

WALES

Preparing for Wales Rare Disease Action Plan

Our key priority is for all the Action Plans to be informed by the views of people living with rare conditions. In Wales we have played a key role in collecting those view points. We collaborated with Genomic Partnership Wales and the Wales Rare Disease Implementation Group to deliver two consultative meetings in November, attended by 47 people between them. These were shared directly with the authors of the Welsh Rare Disease Action Plan.

Working with Wales Gene Park

Working with Wales Gene Park and Genomics Partnership Wales, Genetic Alliance UK continues to hold regular virtual engagement events such as the Genomics Cafes. We now host specific cafe's to engage with younger people (aged 16-25) and spin out cafes such as one held in collaboration with Friends of Cymru Sickle Cell & Thalassaemia, focussing on inherited haematological conditions.

Policy engagement in Wales

HFEA feedback: Genetic Alliance UK statements made the condition 'come alive' and had a 'direct impact' on the committee's decision.

With Emma Hughes, our team member based in Wales Gene Park, on maternity leave, for the second half of the year we split her duties between the Gene Park and our team this year, with our team maintaining connections with the Rare Disease Implementation Group and Genomics Partnership Wales, and providing the secretariat to the Cross Party Group on Rare, Genetic and Undiagnosed Conditions.

Senedd meeting Jan 2022 |

Russell George MS chaired a meeting for Members of the Senedd in January 2022 to reintroduce the topic of rare conditions following the elections. Four MS and their representatives attended the meeting where we broached the topic of diagnosis and care coordination. The attendees heard from an array of speakers including: Martin Williams, member of SWAN UK; Michelle Conway, Rare Disease Nurse Network; Amy Simpson, the CoOrdiNated Care of Rare Diseases (CONCORD) study; and Dr Graham Shortland, SWAN Clinic pilot.

Discussion focussed on next steps for the SWAN Clinic pilot and how it will contribute to improving diagnosis for people with undiagnosed conditions, especially how care would be handled once a diagnosis was received.

RALLY FOR RARE

Candidate pledges doubled since 2016 campaign | 120 in Scotland | 84 in Wales

Between 6 April and 6 May 2021, Genetic Alliance UK ran a pre-election campaign ahead of the Scottish and Welsh elections. We asked our members and supporters of our projects, Rare Disease UK and SWAN UK, to contact their local prospective parliamentary candidates (those standing for election to become MSPs and MSs) to encourage them to pledge their support for people affected by rare, genetic and undiagnosed conditions.

Rally for Rare has helped to raise awareness of rare, genetic and undiagnosed conditions, galvanise our relationships with parliamentarians in the devolved parliaments, and support our efforts to re-establish Cross Party Groups (CPGs) in each nation. The CPGs have been vital in ensuring the voices of people living with rare, genetic and undiagnosed conditions are represented in parliament. The campaign also facilitated the opportunity for our members and supporters to establish closer links with their local representatives.

SCOTLAND

Access to Medicines (Scotland)

Stakeholder consultation | Medicine matters magazine produced

In Scotland, we have been working on the Why Medicines Matter project. In collaboration with Michelle Conway from CRD Consulting (who has supported the project as a Policy Expert Volunteer), we have undertaken a programme of engagement activities to explore how access to medicines in Scotland can improve. The project focused on consultation with three key stakeholder groups; support organisations, clinicians and industry. A summary report of our findings will be shared with the Scottish Government's Rare Disease Team. For this project, it was also important that participants could, in their own words, share why medicines matter to them. To facilitate this we have created a supplementary Why Medicines Matter magazine featuring articles and contributions from key stakeholders which we will use over the course of the next 12 months as part of a social media campaign. You can find Why Medicines Matter at geneticalliance.org.uk/gauk-news/news/why-medicines-matter/.

UK Rare Diseases Framework (Scotland)

We have been working closely with the Scottish Government's Rare Diseases Team to deliver a programme of involvement events for people living with rare conditions in Scotland. Our Policy and Engagement Manager for Scotland has chaired the Scottish Government's Rare Disease Patient Voices Advisory Group and is also a member of the Rare Disease Implementation Board and Short Life Working Group on Care Coordination. The Rare Disease Action Plan for Scotland is expected in summer 2022.

Genetic Alliance UK are represented on the UK Rare Diseases Stakeholder Forum by Natalie Frankish, Policy and Engagement Manager for Scotland.

CROSS PARTY GROUP (CPG) ON RARE, GENETIC AND UNDIAGNOSED CONDITIONS (SCOTLAND)

Following the 2021 Scottish Parliamentary elections, we were quickly able to re-establish the CPG on Rare, Genetic and Undiagnosed Conditions. The group continues to act as a forum for people with lived experience, support groups, clinicians, policy members and elected officials to come together and explore issues relevant to our community. There have been three meetings of the CPG:

CPG: The four priorities of the UK Rare Diseases Framework.

June 2021 | 31 Participants including 2 Members of the Scottish Parliament |

In June we heard from Professor Zosia Miedzybrodzka about the work being undertaken to increase Scotland's genetic testing capabilities, Dr Martina Rodie from Office for Rare Conditions in Glasgow spoke about the need to increase awareness of rare conditions amongst healthcare professionals in NHS Scotland and Dr Catherine McWilliam described the work being undertaken by the Scottish Muscle Network to improve coordination of care and raised the issue of the need to improve the newborn screening programme in Scotland so that people with rare conditions could benefit from treatments available in Scotland. This meeting highlighted the issue of newborn screening for Spinal Muscular Atrophy, which would become a focus for the CPGs work.

Newborn screening.

October 2021 | 29 Participants | 2 parliamentarians

We wrote a letter to the Cabinet Secretary for Health and Social Care to ask for further details of work being undertaken to develop Scotland's Action Plan and to ask for details on Scotland's Newborn Screening Programme, specifically, how decisions on which conditions were included were made. In November we presented our community's view on this topic, followed by Professor Tom Gillingwater and Dr Catherine McWilliam detailed the complex case regarding spinal muscular atrophy in Scotland. The discussion highlighted that it was unclear what role the Scottish Screening Committee had in relation to newborn screening and it was decided that further information should be sought.

CPG: Access to medicines (Scotland).

January 2022 | 41 Participants | 4 Parliamentarians | Presentations on Why Medicines Matter project |

In January, the meeting's theme was priority four of the UK Rare Diseases Framework, improving access to services, treatment and drugs. A presentation was delivered by Michelle Conway (Director of CRD Consulting and Policy Expert Volunteer for Genetic Alliance UK) on the initial findings of the Why Medicines Matter project. We also heard testimony from Kate Learoyd from the National Society for Phenylketonuria (NSPKU) and Michaela Regan from Muscular Dystrophy UK, which highlighted the importance of access to medicines for their communities.

RARE RESOURCES

Rare Resources Scotland

Rare Resources Scotland is a project designed to address information and resource gaps for people living with rare, genetic and undiagnosed conditions in Scotland. Through consultation with members of our community we have identified the information needs of our community and developed a series of information toolkits and guides.

The guides include a toolkit for families with a child or young adult with a rare, genetic or undiagnosed condition, a guide for adults living with a rare condition and a guide about rare conditions for healthcare professionals working in Scotland.

Throughout 2021/22 we have delivered information sessions to the community as part of our Rare Resources offering – this has included sessions on mental health support from Breathing Space and information on financial support for families through Family Fund.

A Rare Resources Information website has been designed to present and disseminate the Rare Resources Scotland toolkit. The webpage has capacity to be expanded to include further resources and can be adapted to include resources from other parts of the UK. You can find out more at geneticalliance.org.uk/information/living-with-a-genetic-condition/rare-resources/.

Discussions are underway to include Rare Resources on NHS Inform, NHS Scotland's health information online resource.

THE VIRTUAL INVOLVEMENT PANEL (SCOTLAND)

Scotland | 80 panel members

In Scotland we have developed a Virtual Involvement Panel for people living with, or caring for someone living with genetic, rare or undiagnosed conditions. The 'VIP' will be at the heart of our work in Scotland and our panellists (our VIPs) will be regularly invited to share their views and experiences to ensure future policy and service planning for rare, genetic and undiagnosed conditions in Scotland meets their needs. VIPs receive regular communication about developments in health and social care services, access to our programme of digital events, opportunities to get involved in our work and invitations to take part in surveys and consultation events to shape policy and services in Scotland.

RESEARCH

Dissemination of CONCORD and mental health findings |one new member group project | two new academic projects

An important aim of our research activities is to support the development of policy by providing reliable and relevant evidence. This year saw a concerted effort to feed the findings of our major study on care coordination (CONCORD) into the national action plans designed to deliver on the commitments of the UK Rare Diseases Framework. Our policy team and members of the study team were able to present and discuss the CONCORD data in several forums and the project is quoted multiple times in the England plan – the first to be published (February 2022). Similarly our 2018 mental health study continues to inform policy work – the Good Diagnosis report uses its findings and those from CONCORD.

We also continue to draw on external scientific studies to support our work. Through a partnership with UCL we were part of a systematic review of evidence around the barriers and facilitators to diagnosis of rare conditions, and we carried out a scoping review of studies about remote (phone/video) healthcare appointments as it became clear that this mode of operation would continue beyond the pandemic. This work is feeding into our policy and further research plans.

Member group research support

We are keen to continue offering research support to our member groups – this can take the form of advice and review, or performing research on a member group's behalf. With Alex TLC we started a new project in March 2022, and began discussions with Ataxia UK for a follow up to their 2020 phase of research.

Mental health support – evaluation

The psychotherapeutic group support that was put in place for our members during the pandemic, through partnering with Rare Minds UK, was extremely helpful according to informal feedback from participants. To properly assess the intervention and build a case for continued support we worked with Manchester Metropolitan University and began an indepth interview study which will report later in 2022.

Evaluation of Rapid Genome Sequencing (rGS) for Critically Ill Children

We are partners in this new NIHR-funded study which is led by researchers at Great Ormond Street Hospital. It will evaluate the introduction of rapid genome sequencing for critically ill children who are in hospital and who urgently need a diagnosis. As with previous projects such as CONCORD, Genetic Alliance UK is in the valuable position of being able to play an active part in the research and at the same time ensure that the opinions of people affected by rare conditions are at the heart of the design and delivery of the study. Since the work began in November 2021 we have developed a survey for parents/carers and established a highly engaged 'public and patient involvement' (PPI) advisory group consisting of parents/carers with a range of relevant experience.

Papers published from research collaborations

Walton H, Simpson A, Ramsay A.I.G. Hunter A, Jones J, Ng PL, Leeson-Beevers K, Bloom L, Kai J, Kokocinska M, Sutcliffe AG, Morris S, Fulop N . Development of models of care coordination for rare conditions: a qualitative study. Orphanet Journal of Rare Disease 17, 49 (2022).

Sanderson SC,Lewis C,Hill M, Peter M, McEntagart M, Gale D, Morris H, Moosajee M, Searle B, Hunter A, Patch C, Chitty LS. Decision-making, attitudes, and understanding among patients and relatives invited to undergo genome sequencing in the 100,000 Genomes Project: A multisite survey study. Genetics in Medicine 24, 61 (2021).

Peter M, Hammond J, Sanderson SC, Gurasashvili J, Hunter A, Searle B, Patch C, Chitty LS, Hill M, Lewis C. Participant experiences of genome sequencing for rare diseases in the 100,000 Genomes Project: a mixed methods study. Eur J Hum Genet 30, 604–610 (2022).

RAPID GENOME SEQUENCING INFOGRAPHIC

Survey for parents and carers of children affected by rare, genetic or undiagnosed conditions

Do you have experience of caring for a child with a rare, genetic or undiagnosed condition?

Can you help to shape our research study by completing a survey?

We would like you to share your views! Everyone who answers the survey will have different views and ideas; they are all important.

What is the survey about?

We are starting a research study to look at how a test called, **"rapid exome sequencing"**, has being offered in the NHS since October 2019. The test is being offered to babies and children who are seriously ill. Those who are offered a test are usually being cared for in a newborn or paediatric intensive care unit. The test can find out if there is a genetic condition which is making them unwell.

This survey is the first step in our two year research study. We are asking you to complete this survey to help us to work out the most important things to look at in our research. It will also help us decide what information would be most helpful to share at the end of the study.

How long will it take?

The survey will take 15-20 minutes to complete. You can complete the survey online, over the phone or on paper. If you would prefer to complete the survey over the phone with a member of the research team, request a paper copy of the survey, or take part in a different language, please get in touch with the research team.

What will the survey ask me to do?

There are three parts to the survey:

Part 1	Part 2	Part 3
Read or watch a short summary of the test	**Read or watch a short summary explaining our research**	**Tell us a little about you**
We will ask you to read or watch a short summary of the test and when it is used. Then there will be some questions asking what you think about the test.	We will ask you to learn about our planned research to evaluate the test. Then there will be some questions about our research.	We will ask some questions about you, such as your age, ethnicity and previous experience of genetic testing.

Who can complete the survey?

You can complete the survey if you are the parent or carer of a child with a rare, genetic or undiagnosed condition. You can also complete the survey if you are a bereaved parent or carer of a child who had a genetic, rare or undiagnosed condition. You must be 18 or over and live in the UK.

Who is organising and funding the survey?

The survey is being carried out by researchers and advisors at Great Ormond Street Hospital, Genetic Alliance UK and Breaking Down Barriers. The survey is funded by the National Institute for Health Research (NIHR202725).

Do I have to take part?

No. It is up to you to decide whether or not to complete the survey. You are free to skip any of the questions in the survey that you would prefer not to answer. You are free to stop filling in the survey at any time before you submit the completed survey. It will not be possible to withdraw your response once you have completed and submitted your answers.

Will my taking part in the survey be kept confidential?

Yes, all information about you will be kept confidential. Some of the questions in the survey will ask you to share your thoughts or provide more detail. It may be helpful for us to use your quotes in an academic publication, conference presentation or report. Quotes will be anonymous – this means we will do what we can to protect your identity so that people reading the quotes will not know who they are from. For example, we will not name people or include the name of any genetic conditions.

What if I find the survey upsetting?

It may be upsetting for you to think about your own experiences with your child. The survey questions are very general and you will not be asked to share specific details about your child and experiences. You can stop at any time and return to the survey later. Because your previous responses will have been saved, you will be able to continue the survey where you left it. If you need support, you can contact the research team and we can put you in touch with someone from your clinical team if you feel this would be helpful, or provide you with details of a patient support group.

Can I find out the results of the survey?

If you wish, you can add your email address to the last page of the survey and we will contact you at the end of the study to send you a short summary of the findings. Your email address will be kept separately from your survey answers.

Who can I contact if I have any questions about the survey?

You can contact the researchers Melissa Hill (Great Ormond Street Hospital) or Amy Hunter (Genetic Alliance UK).

Melissa Hill: RGSevaluation@gosh.nhs.uk/ 020 7405 9200 (ext. 4944)

Amy Hunter: amy.hunter@geneticalliance.org.uk

Download participant information sheet

For more detailed information on participating in the survey, including how we protect your data, please read the participant information sheet by clicking on the link below.

BUILDING OUR ORGANISATION

In 2021/22 we have focused on being responsive to the needs of the diverse community that we serve – everyone affected by a genetic, rare or undiagnosed condition. As the UK's largest alliance of rare and genetic organisations, we have a responsibility to ensure that all voices are heard.

Internally we have carried out a diversity and inclusion review of our internal and external working practices and are committed to taking action to make our organisation fully representative of the community we serve. We are conscious that this is just the start of our journey.

Externally, we have focused on strengthening existing relationships with our members and key stakeholders while also forging new and mending lapsed ones. We have strived to give our members the best possible platform to achieve their aims, including by introducing new member benefits to amplify their voices.

Following the publication of the UK Rare Diseases Framework, we put significant resources into ensuring that there are as many voices of people living with genetic, rare and undiagnosed conditions as possible feeding into the development of the four nations' implementation plans. We did this by empowering our membership to take up formal representative roles, supported by our team where needed, and empowering our broadest networks to feed into the process. We supported all of our representatives and provided a forum for them to exchange information and support each other.

We carried the thread of inclusion through to our work on the UK Rare Diseases Framework to make sure that the voice of our community in this process is diverse and representative. Where we were concerned that there were not enough diverse voices round the table, in terms of both ethnicity and lived experience, we acted as critical friend and raised our concerns proactively with ministers in the UK governments.

We continued to utilise a hybrid approach to working this year, with some staff members co-located in a shared workspace while the majority of the team worked from home. We worked hard to bring the team together virtually and face-to-face where possible to maintain the sense of team spirit that was a key element of our previous office-based environment.

THE COMING YEAR

2022/23 will be an important year for Genetic Alliance UK, as our five-year strategy for 2019-2023 draws to a close and we look to the future. We will evaluate the outcomes of this strategy to learn what has worked well, what has worked less well, and what we would like to do differently in future.

We were joined in July 2022 by our new Chief Executive Louise Fish, who previously led the Tuberous Sclerosis Association, one of the amazing rare disease charities that make up Genetic Alliance UK's membership. Louise's priority will be to lead the development of a new five-year strategy for Genetic Alliance UK setting out how we will continue to drive improvements in treatment and care by speaking with a strong, united voice on behalf of people living with rare, genetic and undiagnosed conditions, their families and the member groups that advocate for them.

Our strategy development will kick off in Autumn 2022 with a listening exercise that puts the individuals, families and organisations we support at its heart. We will shape our five-year strategy through a consultative process with members, potential partners and stakeholders in Spring 2023, so that it is ready for approval by the Genetic Alliance UK Board in Autumn 2023.

We will continue on our journey to make Genetic Alliance UK fully representative of the community we serve by implementing the recommendations resulting from the diversity and inclusion review of our internal and external working practices.

We anticipate that the UK fundraising environment will continue to be challenging for the next few years, due to the global and national impact of the war in Ukraine, rising inflation, and the increasing costs of living. In 2022/23 we will implement the recommendations set out in our recently developed fundraising strategy which sets out how we will diversify and grow our income across corporate partnerships, trusts and foundations, and community fundraising.

In the coming year we will also challenge ourselves to work even more efficiently as a charity, making sure we can invest as much of our time and resources as possible in delivering for our beneficiaries. We will streamline our internal processes and procedures, retender external services to make sure we get good value for money, and investigate the potential to save on office costs by sharing space with one of our member charities.

Most importantly, we will continue to drive the changes that are needed to improve the lives of people living with rare, genetic and undiagnosed conditions and their families. The UK Rare Diseases Framework and the four nations' implementation plans represent a unique opportunity to deliver positive change. We will continue to ensure that the voices of the individuals, families and organisations we support are heard loud and clear in the development of updated implementation plans for each nation in 2022/23. Our goal is to drive demonstrable improvements for our community across diagnosis, awareness of rare conditions, coordination of care, and access to specialist care, treatments and drugs. Working hand-in-hand with the incredible individuals, families and member organisations that we represent, we are stronger together and determined to succeed.

TREASURER'S LETTER AND FINANCIAL REVIEW
31 March 2022

Thank you to all our members and funders. You have ensured that the work outlined in this report could continue at such a challenging time for the people we support.

Review of the financial position

Income

The results for the year are set out in the statement of financial activities on page 42. The total income for year was £713,568 (£939,440 in 2021).

This is a substantial decrease across both restricted and unrestricted income and highlights the continuing challenging fundraising environment. It also should be noted that the previous year saw exceptional support from our supporters in the first year of the pandemic and access to the Government's Job Retention Scheme throughout that period.

A detailed breakdown of income across the two years is included in Note 2 of the Financial Statements.

Expenditure

Total expenditure for the year was £762,424 (£808,907 in 2021).

The decrease in the spending is proportionately smaller than the fall in income, which reflects the focus on the delivery of projects in support of our charitable activities.

The continuing cost control measures taken by the organisation and the focus on delivering impact have led to a substantial fall in support costs during the period as shown in Note 5.

Surplus/Deficit

Our final financial position for the year across all funds is a deficit of £48,856 (surplus of £130,533 in 2021). This total is driven by modest deficits on both restricted and unrestricted funds.

This reflects the exceptional factors that contributed to the surplus in 2021 and the continuing challenging fundraising environment.

Reserves Policy

Total reserves at the end of the financial year are £151,008 (£199,864 in 2021).

This is made up of restricted reserves of £16,351 (£39,134 in 2021) and unrestricted reserves of £134,657 (£160,730 in 2021).

The decrease in restricted reserves is due to slightly higher spending than income on specific projects as detailed in Note 19.

The decrease in unrestricted reserves is due to the inability to maintain unrestricted income levels at prior year levels.

The Trustees have set a target in the medium term to hold unrestricted reserves that equate to approximately 6 months of unrestricted expenditure. Our current level of reserves has decreased over the year and is now approximately 3.5 months by this measure.

The Board and Executive are clear that income generation and cost control measures need to continue to ensure that we can maintain and maximise our impact in the years ahead.

Conclusion

The financial year ended 31 March 2022 was a challenging one for Genetic Alliance UK from an operational and financial perspective, as the second full year of the Covid-19 pandemic.

The Executive Team have demonstrated their ability to lead and manage in a challenging environment, however, it is clear that financial risk still remains and the year ahead will require a continued focus on fundraising and continued cost consciousness.

Genetic Alliance UK starts the period with solid financial foundations. We are pleased that the auditors have endorsed the view that GAUK remains a going concern and we will work to ensure that it continues to be there for all those who need.

D A Ramsden
Treasurer, Board of Trustees

STRUCTURE, GOVERNANCE AND MANAGEMENT

Constitution

Genetic Alliance UK is the trading name for Genetic Alliance UK Ltd, a registered charity and a company limited by guarantee, incorporated on 6 April 2006 and governed by its Memorandum and Articles of Association.

Appointment and training of Trustees

Trustees are elected by the membership. Members co-opted in year and new nominees must be proposed and seconded by a member. Trustee appointments are for three years, after which trustees retire but are eligible for re-election for a further term of three years. New trustees must undertake an induction and are provided with an information pack detailing the charity's work, governance, management policies and procedures, and potential conflicts of interest that may arise.

Governance and organisational management

Trustees met four times in the year ending 31 March 2022, where they reviewed the charity's performance and determined and approved operating plans and budget. Trustees delegate certain powers in connection with the charity's management, remuneration (related to responsibility and market comparisons) and administration to the Finance and Governance (F&G) committee which met four times in the year. The F&G committee provides detailed oversight and advice to the Board of Trustees in relation to financial management, financial viability, risk management and governance. The F&G committee has a minimum of three Trustee members, appointed from and by the Board of Trustees that includes the Treasurer (Chair of the Committee), Chair of Genetic Alliance UK and the Deputy Chair. The Chief Executive and Directors of Genetic Alliance UK are ex officio.

Risk management

Trustees have considered the major risks to which the charity is exposed and have established procedures including a risk register to identify and manage those risks. All risks are reviewed at each meeting of the Board of Trustees and by the F&G committee.

Fundraising

Genetic Alliance UK undertakes most of its fundraising activities in-house but used a consultant solely for the purposes of large grant applications. Genetic Alliance UK is registered with the Fundraising Regulator and adheres to the codes of ethics laid out by the Fundraising Regulator and The Code of Fundraising practice in relation to all fundraising activities. The charity received no complaints about its fundraising practice in this financial year.

REFERENCE AND ADMINISTRATIVE DETAIL

Governing document

The charity is controlled by its governing document, a deed of trust, and constitutes a limited company, limited by guarantee, as defined in the Companies Act 2006.

Registered company number

05772999

Registered charity numbers

1114195 & SC039299

Registered office

Creative Works, 7 Blackhorse Lane, London, E17 6DS

Trustees

Ms Sondra Butterworth (resigned July 2021)
Ms Gloria Clark
Ms Phillippa Farrant
Ms Sara Hunt
Dr Celine Lewis
Mr Neil McClements
Mrs Sue Millman
Mrs Elizabeth Porterfield
Mr David Ramsden
Dr Susan Walsh
Mrs Julie Wooton
Dr Sarah Wynn

Board observer

Robin Nott

Chief Executive Officer

During the period of this report: Lauren Roberts and Nick Meade (appointed as Joint Interim Chief Executives April 2021)

Currently: Louise Fish (joined July 2022)

Management team

Dr Amy Hunter, Nick Meade,
Lauren Roberts (left July 2022)

Auditors

Nyman Libson Paul Chartered Accountants, Regina House, 124 Finchley Road, London, NW3 5JS

Management accountant

Fiona Bevan Financial Management

Bankers

CAF Bank Ltd, 25 Kings Hill Avenue, Kings Hill, West Mailing, Kent, ME19 4JQ

HSBC, Lion House, 25 Islington High Street, London, N1 9LJ

Virgin Money Saving, Jubilee House, Gosforth, Newcastle upon Tyne, NE3 4PL

Website

geneticalliance.org.uk

Facebook

GeneticAllianceUK

Twitter

GeneticAll_UK

Instagram

GeneticAllianceUK

LinkedIn

Genetic Alliance UK

TRUSTEE RESPONSIBILITY

The Trustees (who are also directors of Genetic Alliance UK Ltd for the purposes of company law) are responsible for preparing the Report of the Trustees and the financial statements in accordance with applicable law and United Kingdom Accounting Standards (United Kingdom Generally Accepted Accounting Practice).

Company law requires the Trustees to prepare financial statements for each financial year which give a true and fair view of the state of affairs of the charitable company and of the incoming resources and application of resources, including the income and expenditure, of the charitable company for that period.

In preparing those financial statements, the trustees are required to:

- Select suitable accounting policies and then apply them consistently.
- Observe the methods and principles in the Charity SORP.
- Make judgments and estimates that are reasonable and prudent.
- Prepare the financial statements on the going concern basis unless it is inappropriate to presume that the charitable company will continue in business.

The Trustees are responsible for keeping proper accounting records which disclose with reasonable accuracy at any time the financial position of the charitable company and to enable them to ensure that the financial statements comply with the Companies Act 2006.

They are also responsible for safeguarding the assets of the charitable company and hence for taking reasonable steps for the prevention and detection of fraud and other irregularities.

In so far as the Trustees are aware:

- There is no relevant audit information of which the charitable company's auditors are unaware.
- The Trustees have taken all steps that they ought to have taken to make themselves aware of any relevant audit information and to establish that the auditors are aware of that information.

Auditors

The auditors, Nyman Libson Paul Chartered Accountants, will be proposed for re-appointment at the forthcoming Annual General Meeting.

Report of the Trustees, incorporating a strategic report, approved by order of the Board of Trustees, as the company directors, on 16 August 2022 and signed on the Board's behalf by:

Elizabeth Porterfield
Chair
Board of Trustees

INDEPENDENT AUDITOR'S REPORT TO THE MEMBERS OF GENETIC ALLIANCE UK

Opinion

We have audited the financial statements of Genetic Alliance UK Ltd for the year ended 31 March 2022 set out on pages 42 to 59 which comprise the statement of financial activities, the balance sheet, the cash flow statement and the related notes, including a summary of significant accounting policies. The financial reporting framework that has been applied in their preparation is applicable law and United Kingdom Accounting Standards, including Financial Reporting Standard 102 The Financial Reporting Standard applicable in the UK and Republic of Ireland (United Kingdom Generally Accepted Accounting Practice).

In our opinion the financial statements:

- give a true and fair view of the state of the charitable company's affairs as at 31 March 2022, and of its incoming resources and application of resources, including its income and expenditure, for the year then ended;
- have been properly prepared in accordance with United Kingdom Generally Accepted Accounting Practice; and
- have been prepared in accordance with the requirements of the Companies Act 2006, the Charities and Trustees' Investment (Scotland) Act 2005 and regulations 6 and 8 of the Charities Accounts (Scotland) Regulations 2006 (amended).

Basis for opinion

We conducted our audit in accordance with International Standards on Auditing (UK) (ISAs (UK)) and applicable law. Our responsibilities under those standards are further described in the Auditor's responsibilities for the audit of the financial statements section of our report. We are independent of the charitable company in accordance with the ethical requirements that are relevant to our audit of the financial statements in the UK, including the FRC's Ethical Standard, and we have fulfilled our other ethical responsibilities in accordance with these requirements. We believe that the audit evidence we have obtained is sufficient and appropriate to provide a basis for our opinion.

Conclusions relating to going concern

In auditing the financial statements, we have concluded that the directors' use of the going concern basis of accounting in the preparation of the financial statements is appropriate.

Based on the work we have performed, we have not identified any material uncertainties relating to events or conditions that, individually or collectively, may cast significant doubt on the company's ability to continue as a going concern for a period of at least twelve months from when the financial statements are authorised for issue.

Our responsibilities and the responsibilities of the directors with respect to going concern are described in the relevant sections of this report.

Other information

The trustees are responsible for the other information. The other information comprises the information included in the trustees' annual report, other than the financial statements and our auditor's report thereon. Our opinion on the financial statements does not cover the other information and, except to the extent otherwise explicitly stated in our report, we do not express any form of assurance conclusion thereon.

In connection with our audit of the financial statements, our responsibility is to read the other information and, in doing so, consider whether the other information is materially inconsistent with the financial statements or our knowledge obtained in the audit or otherwise appears to be materially misstated. If we identify such material inconsistencies or apparent material misstatements, we are required to determine whether there is a material misstatement in the financial statements or a material misstatement of the other information. If, based on the work we have performed, we conclude that there is a material misstatement of this other information, we are required to report that fact.

We have nothing to report in this regard.

Opinions on other matters prescribed by the Companies Act 2006

In our opinion, based on the work undertaken in the course of the audit:

- the information given in the trustees' report for the financial year for which the financial statements are prepared is consistent with the financial statements; and
- the directors' report has been prepared in accordance with applicable legal requirements.

Matters on which we are required to report by exception

In the light of our knowledge and understanding of the charitable company and its environment obtained in the course of the audit, we have not identified material misstatements in the Report of the Board of Trustees and Accounts.

We have nothing to report in respect of the following matters in relation to which the Companies Act 2006 and the Charities Accounts (Scotland) Regulations 2006 require us to report to you if, in our opinion:

- adequate accounting records have not been kept, or returns adequate for our audit have not been received from branches not visited by us; or
- the financial statements are not in agreement with the accounting records and returns; or
- certain disclosures of directors' remuneration specified by law are not made; or
- we have not received all the information and explanations we require for our audit; or
- the trustees were not entitled to prepare the financial statements in accordance with the small companies' regime and take advantage of the small companies' exemption in preparing the Report of the Board of Trustees and Accounts.

Responsibilities of trustees

As explained more fully in the trustees' responsibilities statement set out on page 38 the trustees (who are also the directors of the charitable company for the purposes of company law) are responsible for the preparation of the financial statements and for being satisfied that they give a true and fair view, and for such internal control as the trustees determine is necessary to enable the preparation of financial statements that are free from material misstatement, whether due to fraud or error.

In preparing the financial statements, the trustees are responsible for assessing the charitable company's ability to continue as a going concern, disclosing, as applicable, matters related to going concern and using the going concern basis of accounting unless the trustees either intend to liquidate the charitable company or to cease operations, or have no realistic alternative but to do so.

Auditor's responsibilities for the audit of the financial statements

We have been appointed auditor under section 44(1)(c) of the Charities and Trustees Investment (Scotland) Act 2005 and under the Companies Act 2006 and report in accordance with regulations made under those Acts.

Our objectives are to obtain reasonable assurance about whether the financial statements as a whole are free from material misstatement, whether due to fraud or error, and to issue an auditor's report that includes our opinion. Reasonable assurance is a high level of assurance but is not a guarantee that an audit conducted in accordance with ISAs (UK) will always detect a material misstatement when it exists. Misstatements can arise from fraud or error and are considered material if, individually or in the aggregate, they could reasonably be expected to influence the economic decisions of users taken on the basis of these financial statements.

Irregularities, including fraud, are instances of non-compliance with laws and regulations. We design procedures in line with our responsibilities, outlined above, to detect material misstatements in respect of irregularities, including fraud. The extent to which our procedures are capable of detecting irregularities, including fraud is detailed below:

We gained an understanding of the legal and regulatory framework applicable to the company and the industry in which it operates and considered the risk of acts by the charity that were contrary to applicable laws and regulations, including fraud. We designed audit procedures to respond to the risk, recognising that the risk of not detecting a material misstatement due to fraud is higher than the risk of not detecting one resulting

from error, as fraud may involve deliberate concealment by, for example, forgery or intentional misrepresentations, or through collusion.

We focussed on laws and regulations which could give rise to a material misstatement in the financial statements, including, but not limited to, the Companies Act 2006.

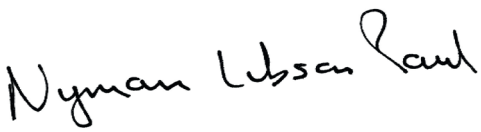
Our tests included agreeing the financial statement disclosures to underlying supporting documentation and enquiries with management. There are inherent limitations in the audit procedures described above and, the further removed non-compliance with laws and regulations is from the events and transactions reflected in the financial statements, the less likely we would become aware of it. We did not identify any key audit matters relating to irregularities, including fraud. As in all our audits, we also addressed the risk of management override of internal controls, including testing journals and evaluating whether there was evidence of bias by the trustees that represented a risk of material misstatement due to fraud.

We also communicated relevant identified laws and regulations and potential fraud risks to all engagement team members and remained alert to any indications of fraud or noncompliance with laws and regulations throughout the audit.

A further description of our responsibilities for the audit of the financial statements is located on the Financial Reporting Council's website at: www.frc.org.uk/auditorsresponsibilities. This description forms part of our auditor's report.

Use of our report

This report is made solely to the charitable company's members, as a body, in accordance with Chapter 3 of Part 16 of the Companies Act 2006 and to the charity's trustees, as a body, in accordance with Regulation 10 of the Charities Accounts (Scotland) Regulations 2006. Our audit work has been undertaken so that we might state to the charitable company's members those matters we are required to state to them in an auditor's report and for no other purpose. To the fullest extent permitted by law, we do not accept or assume responsibility to anyone other than the charitable company and the charitable company's members as a body, for our audit work, for this report, or for the opinions we have formed.



Jennifer Pope (senior statutory auditor)
for and on behalf of

Nyman Libson Paul LLP
Chartered Accountants
Registered Auditors
124 Finchley Road
London
NW3 5JS
Date: 16 August 2022

ACCOUNTS

GENETIC ALLIANCE UK LTD

STATEMENT OF FINANCIAL ACTIVITIES (INCLUDING INCOME AND EXPENDITURE ACCOUNT)

YEAR ENDED 31 MARCH 2022

	Note	Unrestricted Funds £	Restricted Funds £	Total Funds 2022 £	Total Funds 2021 £
Income from:					
Donations and legacies	**2**	383,619	273,835	657,454	933,883
Charitable activities	**3**	56,114	-	56,114	5,557
Total income		439,733	273,835	713,568	939,440
Expenditure on:					
Raising funds	**4**	95,416	-	95,416	105,444
Charitable activities	**5**	368,960	298,048	667,008	703,463
Total expenditure		464,376	298,048	762,424	808,907
Net income/(expenditure)	**8**	(24,643)	(24,213)	(48,856)	130,533
Transfers between funds	**19**	(1,430)	1,430	-	-
Net movement in funds		(26,073)	(22,783)	(48,856)	130,533
Total funds at start of year	**19**	160,730	39,134	199,864	69,331
Total funds at end of year	**19**	134,657	16,351	151,008	199,864

The Charity has no recognised gains or losses other than the results for the year as set out above.

All of the activities of the charity are classed as continuing.

The notes on pages 45 to 59 form part of these financial statements
See note 11 for fund-accounting comparative figures

GENETIC ALLIANCE UK LTD

BALANCE SHEET

AS AT 31 MARCH 2022

Company number: 05772999

	Note	2022 £	2021 £
Fixed assets			
Tangible assets	**12**	992	-
		992	-
Current assets			
Debtors	**13**	66,149	126,373
Cash at bank and in hand		199,282	239,896
		265,431	366,269
Liabilities			
Creditors : amounts falling due within one year	**14**	(115,415)	(166,405)
Net current assets		150,016	199,864
Total assets less current liabilities		150,016	199,864
Net assets		151,008	199,864
FUNDS			
Unrestricted funds			
General funds	**20**	55,109	62,409
Designated funds	**20**	79,548	98,321
Restricted funds	**20**	16,351	39,134
Total funds		151,008	199,864

These financial statements have been prepared in accordance with the special provisions for small companies under Part 15 of the Companies Act 2006.

These financial statements were approved by the Trustees on 16 August 2022 and are signed on their behalf by:

Liz Porterfield
Chair of Trustees

The notes on pages 45 to 59 form part of these financial statements

GENETIC ALLIANCE UK LTD

CASH FLOW STATEMENT

YEAR ENDED 31 MARCH 2022

	Note	2022 £	2021 £
Net cash inflow from operating activities	**16**	(39,336)	127,841
Non-operational cash flows:			
Investing activities			
Payments for tangible fixed assets		(1,278)	-
		(1,278)	-
Net cash inflow/(outflow) for the year	**17**	(40,614)	127,841

Cashflow Restrictions

Charity law prohibits the use of net cash inflows on any endowed or other restricted fund to offset net cash outflows on any fund outside its own objects, except on special authority. In practice, this restriction has not had any effect on cash flows for the year.

The notes on pages 45 to 59 form part of these financial statements

1 Accounting policies

Accounting convention

The financial statements have been prepared in accordance with the historical cost convention (except for investments which have been included at fair value and in accordance with the Statement of Recommended Practice: Accounting and Reporting by Charities preparing their accounts in accordance with the Financial Reporting Standard applicable in the UK and Republic of Ireland (FRS 102) (second edition January 2019) and the Financial Reporting Standard applicable in the United Kingdom and Republic of Ireland (FRS 102) and the *Companies Act 2006* and UK Generally Accepted Practice as it applies from 1 January 2019.

The charity is a public benefit entity as defined under FRS102.

Going concern

The charity faced additional challenges in the year as the economy began to recover from the economic and other consequences of the Covid-19 pandemic, with less government support, and fundraising in particular was impacted. These circumstances resulted in reduced revenue and an unrestricted fund deficit for the year of c £24k compared to a surplus for the previous year of £116k. Unrestricted free reserves (general and designated funds) stand at approximately £135k as at 31 March 2022 and these reserves continue to provide a buffer for the charity.

The Trustees have now reviewed the current circumstances of the Charity and updated budgets, forecasts and projections for the next 12 months have been prepared. Prudent assumptions with regard to income indicate that the Charity will break even for the year ending 31 March 2023 and indications to date are that the fundraising targets included within the budget will be met with the accounts to date showing in excess of 50% of those targets already met 4 months into the 2022/23 year. On the basis of these results and the reserves in hand, adequate liquid resources will continue to be available to fund the activities of the charity for the forseeable future.

Accordingly, the Trustees consider it appropriate for the Charity to continue to adopt the going concern basis in preparing its financial statements.

Income

Income from donations is included in income when these are receivable, except as follows:

I. When donors specify that donations given to the charity must be used in future accounting periods, the income is deferred until those periods;

II. When donors impose conditions which have to be fulfilled before the charity becomes entitled to use such income, the income is deferred until the pre-conditions have been met.

Grants, including government grants are accounted for as receivable and are allocated to Income from Donations and Legacies. Other grants which are received subject to the charity providing a specific level of service are included within Income from Charitable Activities.

Membership income is accounted for as receivable.

Investment income is included on a receivable basis.

Donations in kind comprise donated services where the costs are measurable and the services would otherwise have to be paid for to maintain operational effectiveness.

1 Accounting policies *(continued)*

Expenditure
Expenditure is recognised in the period in which it is incurred. Expenditure includes attributable VAT which cannot be recovered.

Raising funds
Raising funds expenditure include those costs incurred in seeking voluntary contributions and other costs which include the costs of running and participating in fundraising events and collections.

Charitable Activities
Grants awarded are allocated to charitable activities.

Grants awarded are treated as expenditure and a liability in the accounts as soon as they become legal or constructive obligations. In the case of multi-year grant awards, the funding for all years is immediately recognised unless there are conditions which need to be met by the recipient to enable the release of subsequent years' funding.

Governance costs
Governance costs include those costs associated with meeting the constitutional and statutory requirements of the charity and include the audit fees and costs linked to the strategic management of the charity. Governance costs are included within support costs.

Allocation and apportionment costs
Certain expenditure is directly attributable to specific activities and this has been included in those cost categories. Other costs, which are attributable to more than one category, are apportioned across cost categories on the basis of an assessment of workload carried out from time to time.

Overhead support costs have been allocated between fundraising and charitable activities. The apportionment has been allocated on the basis of usage and is analysed in note 6.

Pension costs and other post-retirement benefits
The charity contributes to defined contribution pension schemes. Contributions payable to the charity's pension schemes are charged to the Statement of Financial Activities in the period to which they relate.

Fixed assets
Fixed assets are held at cost less accumulated depreciation. Assets costing less than £500 are not capitalised.
Depreciation is charged on assets at the following rates:
Office equipment - 3 years straight line

Debtors
Debtors are initially measured at the settlement amount after any trade discounts. Subsequently they are measured at the value of the consideration expected to be received.

1 Accounting policies *(continued)*

Cash
Cash balances represent cash and cash equivalents held with a maturity date of less than one year and are included at fair value.

Creditors
Creditors are measured at the settlement amount less any trade discounts.

Fund accounting

Unrestricted funds can be used in accordance with the charitable objects at the discretion of the trustees.

Restricted funds can only be used for particular restricted purposes within the objects of the charity. Restrictions arise when specified by the donor or when funds are raised for particular restricted purposes.

Designated funds form part of unrestricted funds and have been identified as being for particular purposes by the Trustees. They are not restricted and can be transferred to general funds at any time at the discretion of the Trustees.

Further explanation of the nature and purpose of each fund is included in note 19 to the financial statements.

2 Income from donations and legacies

	Unrestricted Funds	Restricted Funds	Total Funds 2022
	£	£	£
Donations from supporters	278,998	150,899	429,897
Legacies received	2,000	-	2,000
Robert Luff Trust	-	20,000	20,000
Wellcome Trust	5,000	-	5,000
University College London	-	5,845	5,845
Edward Gostling	4,689	-	4,689
Hugh Fraser	6,000	-	6,000
Great Ormond Street Hospital	-	10,318	10,318
Grants from Government			
National Lottery funding	-	72,812	72,812
Scottish Government	-	9,961	9,961
Covid Job Retention Scheme fund	6,584	-	6,584
Other grants received	11,308	4,000	15,308
Donations through fundraising:			
Online donations	69,040	-	69,040
	383,619	273,835	657,454

2 Income from donations and legacies *(continued)*

Prior year

	Unrestricted Funds £	Restricted Funds £	Total Funds 2021 £
Donations from supporters	397,745	185,235	582,980
Wellcome Trust	50,590	-	50,590
University College London	-	44,841	44,841
The Corra Foundation	-	36,805	36,805
National Lottery funding	-	25,907	25,907
City Bridge Trust	-	25,700	25,700
Edward Gostling	4,994	-	4,994
Grants from Government			
National Lottery funding	-	20,026	20,026
Scottish Government	-	9,821	9,821
Covid Job Retention Scheme fund	67,202	-	67,202
Other grants received	1,000	-	1,000
Donations through fundraising:			
Community fundraising	7,552	-	7,552
Online donations	55,327	1,138	56,465
	584,410	349,473	933,883

3 Income from: Charitable activities

	Unrestricted Funds £	Restricted Funds £	Total Funds 2022 £
Consultancy work	56,114	-	56,114
	56,114	-	56,114

Prior year

	Unrestricted Funds £	Restricted Funds £	Total Funds 2021 £
Consultancy work	5,557	-	5,557
	5,557	-	5,557

4 Expenditure on: Raising funds

		Total Funds 2022 £	Total Funds 2021 £
Staff costs, including consultancy work		87,483	62,938
Fees		1,438	2,990
Other direct costs		-	44
Support costs	(Note 6)	6,495	39,472
		95,416	105,444

5 Expenditure on Charitable activities

	Direct Costs £	Support Costs (Note 6) £	Total Funds 2022 £
Membership and Engagement	226,973	65,296	292,269
Policy work	173,533	127,939	301,472
Research	34,486	38,781	73,267
	434,992	232,016	667,008

Prior year	Direct Costs £	Support Costs (Note 6) £	Total Funds 2021 £
Membership and Engagement	110,002	114,597	224,599
Policy work	122,778	194,119	316,897
Research	71,233	90,734	161,967
	304,013	399,450	703,463

£176,390 of the above support costs in notes 4 and 5 and £276,026 of direct costs relate to unrestricted funds spent during the year. £62,121 of support costs and £235,927 of direct costs relates to restricted funds spent during the year.

£244,779 of the above support costs in notes 4 and 5 and £229,278 of direct costs relate to unrestricted funds spent during the prior year. £194,143 of support costs and £140,707 of direct costs relates to restricted funds spent during the prior year.

6 Support costs

Support costs are allocated between raising funds and charitable activities on the basis of usage.
Support costs, included in notes 4 & 5, are as follows:

		Raising Funds £	Charitable Activities £	Total 2022 £
Staff costs		-	127,654	127,654
Communications		352	2,947	3,299
Office and admin costs		6,143	51,373	57,516
Travel		-	2,369	2,369
Finance costs		-	772	772
Professional fees		-	35,801	35,801
Governance costs	(Note 7)	-	11,100	11,100
		6,495	232,016	238,511

Prior year		Raising Funds £	Charitable Activities £	Total 2021 £
Staff costs		32,455	271,435	303,890
Communications		1,637	13,691	15,328
Office and admin costs		5,380	44,999	50,379
Travel		-	231	231
Finance costs		-	889	889
Professional fees		-	58,005	58,005
Governance costs	(Note 7)	-	10,200	10,200
		39,472	399,450	438,922

7 Governance costs

	Total Funds 2022 £	Total Funds 2021 £
Auditor's fees - for audit services	8,100	7,200
Accounts review	3,000	3,000
	11,100	10,200

8 Net income/(expenditure) for the year

This is stated after charging:

		2022 £	2021 £
Auditor's remuneration	- for audit services	8,100	7,200
Trustees' travel expenses (2 trustees)		884	-

No Trustee received any remuneration during the year.

9 Staff costs and numbers

The aggregate payroll costs were:

	2022 £	2021 £
Wages & salaries	446,757	483,459
Social security costs	41,839	47,496
Pension contributions	25,283	33,650
Redundancy costs	8,338	33,901
	522,217	598,506

The number of employees whose employee benefits exceeded £60,000 during the year were:

	2022	2021
Between £60,001 and £70,000	1	-
Between £70,001 and £80,000	-	1

The average weekly number of employees during the year was 17 (2021: 17), calculated on the basis of average headcount. The total employment benefits received by key management personnel including employer's national insurance and employer's pension contributions were £181,356 (2021: £234,821).

10 Taxation

The charity is exempt from corporation tax on its charitable activities.

11 Statement of Financial Activities comparative figures

For the year ended 31 March 2021	Unrestricted Funds £	Restricted Funds £	Total Funds 2021 £
Income from:			
Donations and legacies	584,410	349,473	933,883
Charitable activities	5,557	-	5,557
Total income	589,967	349,473	939,440
Expenditure on:			
Raising funds	105,444	-	105,444
Charitable activities	368,613	334,850	703,463
Total expenditure	474,057	334,850	808,907
Net income/(expenditure)	115,910	14,623	130,533
Transfers between funds	42	(42)	-
Net movement in funds	115,952	14,581	130,533
Total funds at start of year	44,778	24,553	69,331
Total funds at end of year	160,730	39,134	199,864

12 Tangible fixed assets

	Office equipment	Total
	£	£
Cost or valuation		
At 1 April 2021	-	-
Additions	1,278	1,278
At 31 March 2022	1,278	1,278
Depreciation		
At 1 April 2021	-	-
Charge for the year	286	286
At 31 March 2022	286	286
Net book value		
At 31 March 2022	992	992
At 31 March 2021	-	-

13 Debtors

	2022	2021
	£	£
Due in less than one year:		
Trade debtors	47,136	125,729
Prepayments and accrued income	17,813	-
Other debtors	1,200	644
	66,149	126,373

14 Creditors: amounts falling due within one year

	2022	2021
	£	£
Trade creditors	6,165	7,551
Social security and other taxes	20,617	11,248
Other creditors	4,320	3,446
Accruals and deferred income	84,313	144,160
	115,415	166,405

15 Operating lease commitments

At 31 March 2022, the charity had future minimum lease payments under non-cancellable operating leases as follows:

	Equipment	
	2022	2021
	£	£
Expiry Date:		
Not later than one year:	-	5,171
	-	5,171

16 Reconciliation of net movement in funds to net cash inflow from operating activities

	2022	2021
	£	£
Statement of Financial Activities: Net movement in funds	(48,856)	130,533
Depreciation	286	-
(Decrease)/increase in creditors: current liabilities	(50,990)	(23,174)
Decrease / (increase) in debtors	60,224	20,482
Net cash inflow/(outflow) from operating activities	(39,336)	127,841

17 Analysis of changes in cash during the year

	2022	2021	**Change**
	£	£	**£**
Cash at bank and in hand	199,282	239,896	(40,614)

	2021	2020	**Change**
	£	£	**£**
Cash at bank and in hand	239,896	112,055	127,841

18 Analysis of changes in net debt

	1 April 2021 £	Cashflow Movements £	31 March 2022 £
Cash at bank and in hand	239,896	(40,614)	199,282
	239,896	(40,614)	199,282

Prior year	1 April 2020 £	Cashflow Movements £	31 March 2021 £
Cash at bank and in hand	112,055	127,841	239,896
	112,055	127,841	239,896

19 Movement in Funds

For the year ended 31 March 2022

	At 1 April 2021 £	Income £	Expenditure £	Transfers £	At 31 March 2022 £
Restricted funds					
Scotland Boost	1,309	5,961	(7,999)	729	-
Action for Access	18,401	-	(18,401)	-	-
Concord	-	5,845	(5,142)	-	703
RD PSPs	5,442	-	(6,143)	701	-
SWAN UK: Dads' Summit	-	4,749	(4,749)	-	-
SWAN UK Wales	9,928	54,286	(46,882)	-	17,332
Building Rare Resilience	-	20,026	(20,026)	-	-
Talking about Gene Therapy	4,054	-	(4,054)	-	-
Patient survey	-	8,066	(8,066)	-	-
Rare Resources	-	7,054	(7,054)	-	-
Building Rare Resilience - support	-	59,232	(59,232)	-	-
Robert Luff trust	-	20,000	(20,000)	-	-
Alex TLC X-ALD study	-	-	(547)	-	(547)
Rapid Genome Sequencing	-	10,318	(11,455)	-	(1,137)
Rare Disease Day	-	78,298	(78,298)	-	-
	39,134	273,835	(298,048)	1,430	16,351
Unrestricted funds					
General funds	62,409	130,096	(170,833)	33,437	55,109
Designated funds					
Rare Disease Day	34,867	-	-	(34,867)	-
Rare Disease UK General Donations	63,454	250,241	(248,196)	-	65,499
SWAN UK: General Donations	-	59,396	(45,347)	-	14,049
	160,730	439,733	(464,376)	(1,430)	134,657
Total funds	199,864	713,568	(762,424)	-	151,008

Fund descriptions
Restricted funds

Scotland Boost - funds received for the dissemination of a toolkit for families affected by genetic, rare and undiagnosed conditions.

Action for Access - funds received to help with the dissemination of the messages in the Action for Access report.

Concord - funds received for research on coordination of care in the UK and dissemination of findings.

19 Movement in funds *(continued)*

RD PSPs - funds received for establishing, and dissemination of, research priorities for mitochondrial disease.

SWAN UK: Dads' Summit - funds received for outreach for dads of children affected by undiagnosed genetic conditions.

SWAN UK Wales - funds received for the development of a support network in Wales for families with children affected by undiagnosed genetic conditions.

Building Rare Resilience - This was the second instalment of restricted funding from the National Lottery Covid-19 emergency fund for the Building Rare Resilience project.

Talking about Gene Therapy - funds are restricted to fund workshops with people living with rare and genetic conditions and report on the outcomes.

Patient survey - funds are restricted to implementing and analysing a survey on individuals' experiences of living with rare and genetic condition and disseminating the findings.

Rare Resources - funds are restricted to the development and dissemination of a toolkit for families affected by genetic, rare and undiagnosed conditions.

Building Rare Resilience - support- funds are restricted to supporting our members with the challenges associated with Covid-19.

Robert Luff trust - funds received from the trust for undertaking research.

Alex TLC X-ALD study - funds received to participate in the Alex TLC X-ALD study**.**

Rapid Genome Sequencing - funds received for research into the Rapid genome sequencing for the diagnosis of critically ill children in the NHS Genomic Medicine service: Ensuring an equitable and effective parent and patient-centred service.

Rare Disease Day - funds received to help with the costs of putting on our annual Rare Disease Day**.**

Genomic Data - funds were received to complete work to understand the attitudes of people living with genetic, rare and undiagnosed conditions and storage of their data.

Covid-19 - funds received to help with the challenges associated with Covid-19.

Designated funds

Rare Disease Day - funds were set aside by the trustees in previous years to support awareness of rare conditions. During the current year, it was decided that there was no longer any need for the funds to be separated, so the balance remaining has been transferred back to general funds.

SWAN UK General Donations - the trustees have set aside these funds received as general donations to SWAN UK, to ensure they are spent on activities within the SWAN UK community.

Rare Disease UK General Donations - national campaign for people with rare diseases and all who support them, providing a united voice for the rare disease community by capturing the experiences of patients and families. Rare Disease UK is focused on making sure the new UK Rare Diseases Framework is as successful as possible, and to ensure that patients and families living with rare conditions have equitable access to high quality services, treatment and support.

19 Movement in funds *(continued)*

For the year ended 31 March 2021

	At 1 April 2020	Income	Expenditure	Transfers	At 31 March 2021
	£	£	£	£	£
Restricted funds					
Rare Disease Day 2018	28	-	(28)	-	-
Scotland Boost	-	9,821	(8,512)	-	1,309
Action for Access	13,047	10,000	(4,646)	-	18,401
Concord	1,061	44,841	(45,902)	-	-
RD PSPs	6,881	-	(1,439)	-	5,442
SWAN UK: Dads' Summit	-	6,364	(6,364)	-	-
SWAN UK Wales	-	25,907	(15,979)	-	9,928
Genomic Data	42	-	-	(42)	-
Building Rare Resilience	-	20,026	(20,026)	-	-
Talking about Gene Therapy	3,494	10,000	(9,440)	-	4,054
Patient survey	-	38,600	(38,600)	-	-
Rare Resources	-	2,846	(2,846)	-	-
Covid-19	-	158,505	(158,505)	-	-
Rare Disease Day	-	22,563	(22,563)	-	-
	24,553	349,473	(334,850)	(42)	39,134
Unrestricted funds					
General funds	44,778	182,069	(162,607)	(1,831)	62,409
Designated funds					
Rare Disease Day	-	60,000	(25,133)	-	34,867
Rare Disease UK General Donations	-	290,748	(227,294)	-	63,454
SWAN UK: General Donations	-	57,150	(59,023)	1,873	-
	44,778	589,967	(474,057)	42	160,730
Total funds	69,331	939,440	(808,907)	-	199,864

20 Analysis of net assets between funds

As at 31 March 2022	Restricted Funds	Unrestricted Designated Funds	Unrestricted General Funds	Total
	£	£	£	£
Tangible fixed assets	-	-	992	992
Bank and cash	16,351	79,548	103,383	199,282
Other net assets	-	-	(49,266)	(49,266)
	16,351	79,548	55,109	151,008

20 Analysis of net assets between funds (continued)

As at 31 March 2021	Restricted Funds £	Unrestricted Designated Funds £	Unrestricted General Funds £	Total £
Bank and cash	39,134	98,321	102,441	239,896
Other net assets	-	-	(40,032)	(40,032)
	39,134	98,321	62,409	199,864

21 Related party transactions

There are no transactions with trustees or other related parties other than those disclosed as required by the SORP elsewhere in the financial statements.

ACKNOWLEDGEMENTS

Thank you to all the individuals and organisations who have supported our work this year. We are so grateful to everyone who has worked with us, volunteered or fundraised for us, or supported our work in other ways.

We would also like to thank all our funders who have given grants, sponsorship or donations to support our work this year. These organisations are listed here.

Thank you to the SWAN UK families who have given us permission to use photographs of their beautiful children in this report.

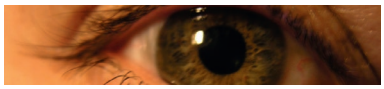
Albireo Pharma
Alexion Pharma
Amicus Therapeutics
Barratt Charity Foundation
BioCryst UK Limited
Biogen Idec Limited
Catherine Cookson Charitable Trust
Chiesi Ltd
CRD Consulting
Edward Gostling Foundation
Gilead Sciences Ltd
HFEA
HRA Pharma Rare Diseases
Hugh Fraser Foundation
Incyte Biosciences UK Ltd
Intent Health
Ionis Pharmaceuticals
Joicey Trust
Sir Jules Thorn Charitable Trust
Kyowa Kirin Ltd
Marjory Boddy Charitable Trust
Medpace UK Ltd
Melton Mowbray Foundation
National Lottery Trust
Novartis Gene Therapies
Novartis Pharmaceuticals UK Ltd
Oakdale Trust
Orchard Therapeutics
Persimmon Charitable Foundation
PF Charitable Trust
Pfizer Limited
PTC Therapeutics
Great Ormond Street
Rhythm Pharmaceuticals
Robert Luff Trust
Roche Products Limited
Russell Trust
Sanofi Genzyme
Scottish Government
SOBI UK Ltd
Takeda UK Limited
Triducive Partners Ltd
UCB Pharma Limited
UCL Research
Vertex Pharmaceuticals
Wellcome Trust

OUR MEMBERS

The Aarskog Foundation

Action Duchenne

Action FCS



Action on Gilbert's Syndrome

Addison's Disease Self-Help Group

Adrenal Hyperplasia Network

Advocacy for Neuroacanthocytosis Patients

Albinism Fellowship UK Ireland

Alex, The Leukodystrophy Charity

ALK Positive Lung Cancer (UK)

Alkaptonuria Society

Alpha-1 Awareness UK

Alpha-1 UK Support Group

Alport UK

Alström Syndrome UK

Amy and Friends

ANE International

Angelman UK

Aniridia Network

Annabelle's Challenge

Anorchidism Support Group

Antenatal Results and Choices

Archangel Mld Trust

Association For Glycogen Storage Diseases UK

Ataxia UK

Ataxia-Telangiectasia Society

Bardet-Biedl Syndrome UK

Barth Syndrome UK

Batten Disease Family Association

Beat SCAD

Beckwith Wiedemann Support Group

Beckwith-Wiedemann Support Group UK

Behçet's UK

BRCA Umbrella

British Heart Foundation

Cadasil Support UK

Cambridge Rare Disease Network

Cardiomyopathy UK

Cavernoma Alliance UK

CdLS Foundation UK and Ireland

CGD Society

CHAMP1 Patient Support Group

Charcot-Marie-Tooth UK

Childhood Tumour Trust

Children's Liver Disease Foundation

Children's Mitochondrial Disease Network

Children's Health Scotland

CML Support Group

Congenital Adrenal Hyperplasia Support Group

Costello Support Group International

Cri Du Chat Syndrome Support Group

Cure and Action for Tay-Sachs Foundation

Cure Myotonic Dystrophy UK Charity

Cystic Fibrosis Trust

Cystinosis Foundation UK

DC Action

DDDC3G Renal Support Group UK

Dercums Disease and Rare Disease Foundation UK

Diamond Blackfan Anaemia Support Group UK

Dravet Syndrome UK

Duchenne Family Support Group

East Lancashire Community Genetics Service

East London Branch Sickle Cell Society

Ehlers-Danlos Society

EHLERS-DANLOS SUPPORT UK

ELIJAH'S HOPE

FANCONI HOPE CHARITABLE TRUST

Fap Gene Support Group

Fibromuscular Dysplasia Society

Fibrous Dysplasia Support Society UK

Fight for Sight

Findacure

FND Hope UK

FSH Muscular Dystrophy Support Group

Fuchsfriends UK

Funny Lumps

GATA2 Deficiency Support Group

GENE People UK

George Pantziarka TP53 Trust

GIST Support UK

Gorlin Syndrome Group

Hadiza Foundation

HAE UK

Haemochromatosis UK

Haemophilia Society

Headlines Craniofacial Support

Hereditary Spastic Paraplegia Group

Hope for Hasti

HPS Network UK

Huntington's Disease Association UK

Hypermobility Syndromes Association

Ichthyosis Support Group

IIH UK

Inherited Prion Disease Support Group

Jeune Syndrome Foundation

Jnetics

KIF1A.ORG

Kleefstra Syndrome

Krabbe UK

Leber's Hereditary Optic Neuropathy Society

Lipodystrophy UK

Local Families With Bleeding Disorders

Lynch Syndrome UK

Macular Society

Making It Better - The Daniel Courtney Trust

Manchester Sickle Cell and Thalassaemia Centre

Marfan Trust

Mast Cell Action

Max Appeal

MEBO Research

Medics 4 Rare Diseases

Metabolic Support UK

Moebius Research Trust

Motor Neurone Disease Association

Mowat-Wilson Syndrome Support Group

MPGN/DDD Support Group

Muscle Help Foundation

Muscular Dystrophy UK

Myotubular Trust

Naitbabies

Narcolepsy UK

National Deaf Children's Society

NCBRS Worldwide Foundation

Nemaline Myopathy Support Group

Nerve Tumours UK

Neuroendocrine Cancer UK

NF2 BioSolutions

Niemann-Pick UK

NLRP12.COM

Noonan Syndrome Association

Northern Ireland
Rare Disease Partnership

Ocumel UK

Osteopetrosis Support Trust

Pans Pandas UK

Parathyroid UK

Pemphigus Vulgaris Network

Pitt Hopkins UK

PNH Support Group

Poland Syndrome
Support & Network

Prader-Willi Syndrome
Association UK

Primary Ciliary Dyskinesia Family
Support Group

Primary
Immunodeficiency UK

PSC Support Group

Pseudoxanthoma Elasticum Support Group

PTEN UK and Ireland Patient Group

Rare Autoinflammatory Conditions Community - UK

Rare Dementia Support

RETT UK

Reverse RETT

RING20 Research and Support UK

SADS UK

Salivary Gland Cancer UK

Sarcoidosis UK

Schinzel-Giedion Syndrome Foundation

Scleroderma And Raynaud's UK

Sense Usher Service

Shwachman-Diamond Support UK

SOFT UK

Special Needs Jungle

Spinal Muscular Atrophy UK

Spotlight YOPD

Stargardt's Connected

Stickler Syndrome UK

Stiff Person Syndrome

Syngap Research Fund

Thalidomide Society

The Aortic Dissection Charitable Trust

The Aplastic Anaemia Trust

The British Porphyria Association

The Children's Hyperinsulinism Charity

The Fragile X Society

The Gauchers Association

The Lily Foundation

The Maddi Foundation

The MPS Society

The Smith-Magenis Syndrome Foundation UK

The UK Mastocytosis Support Group

Thyroid UK

Timothy Syndrome Alliance

TreatSMA

TRPS Support Group UK

Tuberous Sclerosis Association

Turner Syndrome Support Society UK

UK LGLL

UK Thalassaemia Society

UKPIPS

Unique – Understanding Chromosome Disorders

Urddad Foundation

Vasculitis UK

VHL UK/Ireland

Williams Syndrome Foundation Limited

Wilson's Disease Support Group UK

Wolfram Syndrome UK

Womb Cancer Support UK

Worster-Drought Syndrome Support Group

XLP Research Trust

XP Support Group

Yellow Brick Road Project Inc

Registered company number: 05772999
Registered charity numbers: 1114195 and SC039299
Registered office: Creative Works, 7 Blackhorse Lane, London, E17 6DS