2023/2024

IMPACT REPORT

GENETIC ALLIANCE UK

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Background

Genetic Alliance UK is an alliance of over 200 charities and support groups. We have a thirty-year track record of working together to improve the lives of people in the UK living with lifelong and complex genetic, rare and undiagnosed conditions.

- Rare conditions are individually rare but collectively common, with over 3.5 million people in the UK living with a rare condition
- A rare condition affects fewer than one in 2,000 people.
- One in 17 people are affected by a rare condition at some point in their lives
- There are around 7,000 rare conditions, with new conditions regularly identified through scientific progress
- Seven in 10 rare conditions affect children
- More than 3 out of 10 children with a rare condition die before their fifth birthday

People living with genetic, rare and undiagnosed conditions and their families 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.

Message from our Chair and Chief Executive

Genetic Alliance UK is an alliance of over 200 charities and support groups. The end of the membership and financial year 2023/24 is an opportunity for us to reflect on the past 12 months and acknowledge the progress we've made together.

Rare conditions are individually rare, but collectively common, affecting 3.5 million people in the UK. Reflecting on what we've achieved together in the past year demonstrates that we are stronger when we collaborate and work together.

We're proud to amplify the diverse voices of charities and support groups representing people living with a wide range of genetic, rare and undiagnosed conditions. Our success belongs to our members: we could not do what we do without each and every one of them.

In addition to highlighting our shared achievements in 2023/24 under the leadership of outgoing Chief Executive Louise Fish, we welcome our new Chief Executive Mark Flannagan who joins us in September 2024. We are looking forward to continuing to work with and on behalf of our community in the year ahead.

Elizabeth Porterfield,
Chair, Genetic Alliance UK

Louise Fish,
Chief Executive Officer, Genetic Alliance UK

Our priorities for 2023/24

During 2023/2024 Genetic Alliance UK continued to deliver on our strategic priorities:

Growing our alliance and supporting our members

Championing timely diagnosis and coordinated care for people living with rare conditions

Providing a strong voice for people living with genetic, rare and undiagnosed conditions

Ensuring scientific breakthroughs in genomics drive better research and services

Promoting opportunities to expand screening in line with international best practice

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. Rare Disease UK is the official UK organiser of Rare Disease Day.
- SWAN UK, the only dedicated support network in the UK for families affected by a syndrome without a name, a genetic condition so rare that it often goes undiagnosed.

Our new strategy

During 2023/24 we developed a new five-year strategy for 2024-2029.

Our purpose (why our alliance exists) is to work together to improve the lives of 3.5 million people in the UK living with lifelong and complex genetic and rare conditions.

Our aim (what we will do) is to make sure the ambitions in the UK Rare Disease Framework that matter most to people living with genetic, rare and undiagnosed conditions drive action across the four nations.

Our objectives (how we will do it) are:

- Influencing national policy and provision where it matters most by:
  - Championing timely diagnosis and better coordinated care and treatment for people living with genetic, rare and undiagnosed conditions.
  - Making sure an understanding of what is most important to people living with genetic, rare and undiagnosed conditions shapes research, policy, regulation, practice and the development of new medicines and therapies.
  - Ensuring scientific breakthroughs in genomics reach people living with genetic conditions in a timely fashion through research programmes and routine diagnostic and clinical services.
  - Promoting opportunities to expand newborn and population screening for genetic, rare and undiagnosed conditions in line with international best practice.
- Making sure our influencing drives action across both policy and practice.
- Tracking how people with genetic, rare and undiagnosed conditions are affected by this action across the four nations.

Our critical success factors (how we are going to get there) are:

- Building a robust, resilient and flourishing alliance in terms of members, income, staff and trustees.
- Working collaboratively with key stakeholders such as umbrella groups for genetic, rare and undiagnosed conditions, healthcare professional bodies and industry.
- Developing an effective influencing strategy to drive action related to the ambitions in the UK Rare Disease Framework that matter most.
- Monitoring and evaluating implementation of the UK Rare Disease Framework.

Our enablers (the things that will make it possible) to deliver this strategy are:

- Putting our members and the people living with genetic, rare and undiagnosed conditions they support at the heart of everything we do: listening to their experience, learning from their expertise, helping members share knowledge with each other, and ensuring our voices are stronger when we work together.
- Professionalising our approach to membership, income generation, corporate services and communications.
- Building on the strengths of our well-regarded policy and research teams.
- Investing in our staff and supporting their training and development.
- Recruiting and retaining trustees with the skills, knowledge and diverse perspectives needed to scrutinise and shape our work.

Our values (the things we believe are important) are to be people-centred, collaborative, inclusive, influential, evidence-led, knowledge-generating and independent.

The development of our five-year strategy for 2024 to 2029 involved consultation with members, supporters and funders. The strategy was launched at the charity's Annual General Meeting in Autumn 2023. Each financial year we will develop a business plan and budget setting out our work for the coming year based on the aims and objectives in our strategy.

1. Growing our alliance and supporting our members

Building a growing and diverse alliance

During 2023/24 our membership increased. Our members range in size from a small number of household names like the Cystic Fibrosis Trust and DEBRA, through to a large number of specialist support groups led by parents of children with rare conditions, and affected adults, volunteering alongside their caring and work responsibilities. We believe our voices are stronger when we work together. We keep membership fees as low as possible to maximise the number of organisations working together to improve the lives of people living with genetic, rare and undiagnosed conditions.

New members and improved opportunities for engagement

We welcomed 19 new member organisations into Genetic Alliance UK during 2023/24. These are:

- ACTA2 Alliance
- Arthur's Quest
- Cure DHDDS
- CUREUsher
- The Down's Syndrome Association
- The Epilepsy Society
- FAR - Foundation for ARID1B Research
- The Gary Woodward Dyskeratosis Congenita Trust
- Galactosaemia Support Group
- GRACE Project
- Little Hearts Matter
- Myaware
- The Neurological Alliance
- Pallister-Killian Syndrome UK (PKSUK)
- PCD Research - Primary Ciliary Dyskinesia
- Rareminds
- RareQoL Ltd
- Retina UK
- Thrombosis UK

During 2023/24 we held 12 monthly Member Briefings to which we invited senior stakeholders to speak directly to our member charities and support groups. Member Briefings were well-attended with over 150 individuals joining at least one briefing and an average of 24 people attending each session.

We also held 12 monthly Member Brews where we brought our members together in a closed session to discuss common challenges and opportunities and share learning with one another. Member Brews were a popular way for members to work together, with over 100 individuals joining at least one brew and an average of 19 people attending each session.

Every month we are creating more opportunities for member organisations to amplify their voice and extend their reach. Examples include presentation slots at Member Briefings and Member Brews, increased promotion of awareness days and weeks, and improved engagement through social media channels and member networks.

Our new Genetic Alliance UK member social media takeovers were a huge success with over 20 member organisations taking part. We also hosted 39 takeovers on our Rare Disease UK Instagram, helping to increase the online reach of our members and individuals living with rare, genetic and undiagnosed conditions in the wider community.

Our new website

In 2020, Genetic Alliance UK carried out the Rare Experience survey to better understand the experiences and preferences of people affected by rare conditions. This survey highlighted the need for timely signposting to relevant patient organisations, as well as the need for easy to access relevant information from a credible source.

In 2023/24 we launched a new website to help our members meet these challenges. We have been working hard to turn geneticalliance.org.uk into a leading information and signposting hub for the UK's rare disease community, which will help raise awareness of our members and their amazing work, whilst also making it easier for the community to access relevant and credible information.

The new website will enable prospective members to join Genetic Alliance UK, engage with our resources and sign up to attend our virtual member meetings. It will also signpost our members to wider sources of reliable specialist support for the rare and genetic conditions sector in the UK, and make it easier for our members, industry partners and policy-makers to find our extensive library of policy reports and research papers on issues of shared concern for the sector.

We launched our new website in February 2024 thanks to the generous support of our industry partners. We continue to add updates, information resources and new content to benefit our members, stakeholders and the wider community.

2. Championing timely diagnosis and coordinated care for people living with rare conditions

Driving progress on UK Rare Diseases Framework

The UK Rare Diseases Framework was published in January 2021. This document outlines the governments' priorities for improving care for people living with rare conditions to be implemented over five years (2021-2026). In 2023/24 Genetic Alliance UK continued to work with its members to support the delivery of the UK Rare Diseases Framework and action plans. The Genetic Alliance UK team holds positions across the delivery groups responsible for the Framework and has been recognised in three of the four action plans for our role in their delivery. Our Rare Disease UK Patient Empowerment Group Recommendations Paper has been considered in meetings of all four nations in the development and implementation of their action plans.

In England, we worked with the Department of Health and Social Care to re-establish the England Rare Disease Action Plan Patient Advisory Group which directly fed into the development of the 2024 Action Plan. In Scotland, we are responsible for supporting the Scottish Government's patient involvement work for the Scottish Rare Disease Action Plan. In Wales, we sit on the Rare Disease Implementation Network (RDIN) which has responsibility for implementing the Welsh Rare Disease Action Plan and recently joined the newly established RDIN Leadership Group.

Working in partnership to deliver the Rare Disease Research UK Platform

A major new £14 million investment by the National Institute of Health Research (NIHR) and Medical Research Council (MRC) into rare condition research launched in 2023. They have invested in 11 'nodes' addressing different challenges - some are focused on groups of conditions or disease 'pathways', while others are cross-cutting such as developing innovative trial design to take account of small populations and to reduce trial costs. A coordinating 'hub' function, led by Newcastle University in partnership with Genetic Alliance UK and Newcastle upon Tyne Hospitals NHS Foundation Trust, will support the nodes to achieve impact from their work and promote good practice in involving people with lived experience.

Additionally, Genetic Alliance UK is providing direct support to two of the nodes, to facilitate the involvement of people with lived experience: CAPTIVATE (focussed on innovative trial design) and UPNAT (progressing the development of a new class of therapies for rare genetic conditions).

Maximising opportunities to raise awareness on Rare Disease Day 2024

This year was a leap year so Rare Disease Day fell on 29 February, a date originally chosen for the annual international awareness day due to its rarity. On Thursday 29 February 2024, Rare Disease Day was celebrated in over 85 countries to raise awareness and generate change for the 300 million people worldwide living with a rare condition, and their families and carers.

Every year the theme is chosen by EURORDIS – Rare Diseases Europe, in collaboration with their patient organisation members. As the official organiser of Rare Disease Day in the UK through our campaign Rare Disease UK, Genetic Alliance UK coordinated closely with EURORDIS, our member organisations and relevant stakeholders to deliver a wide range of activities to express our international solidarity and highlight the priorities of the UK's rare community.

Central source of data about rare conditions

On Rare Disease Day 2024 we raised awareness of facts and figures about rare diseases in the UK with NHS staff, healthcare professionals, people living with rare disease and the public. We developed and published a central source of data about rare conditions in the UK which is hosted on the new Genetic Alliance UK website. These facts and figures provide an easy and impactful way to raise awareness and understanding of the challenges of living with a rare, genetic or undiagnosed condition in the UK.

Policy report to drive better understanding of data

Our Stats Behind the Stories policy report argues that we need to segment and better understand UK data about who has rare conditions, and which rare conditions they have, so that the NHS can provide the right services and support. The report includes case studies from Hereditary Brain Aneurysm Support, Superficial Siderosis Research Alliance, Better Together for Healthy Marrow Alliance and the Neurological Alliance to bring a human face to the facts and figures discussed in it. We would like to thank the member organisations who shared these vital stories with us.

Dissemination of the report has been a key focus since Rare Disease Day. We have met with the England Rare Disease Action Plan Delivery Group, the UK Rare Disease Framework Stakeholder Forum, and the British Society of Genetic Medicine. We plan to meet with the Wales Rare Disease Implementation Network and the UK Rare Disease Framework delivery partners. We are discussing the findings of the report and exploring a pragmatic approach to gaining more understanding of how rare conditions affect the UK population and how we can develop future solutions.

Factsheets to raise awareness of key facts and figures

We produced six clear and simple factsheets on key areas relating to rare conditions which are available on our new website, and we encouraged our members and stakeholders to share and use these factsheets in their own work. The six factsheets focus on: facts about rare, genetic and undiagnosed conditions; UK Rare Diseases Framework and Action Plans; diagnosis of rare conditions; coordinating care for rare conditions; newborn screening for rare conditions; and access to rare disease medicines in the UK.

Raising awareness through the media

We partnered with ITN Business to develop a high-quality online programme that can be widely shared by interested organisations and stakeholders. Our programme, 'Rare Conditions: The Stories Behind the Stats' went live on Rare Disease Day 2024 and uses a high-quality animation developed by the ITN graphics team to bring the facts and figures highlighted throughout this year's campaign to life. We also supported the Rare Diseases Campaign that launched within The Guardian and online through an exclusive piece from our Director of Policy, Nick Meade, exploring how to manage rare conditions in the UK effectively.

Raising awareness at Parliamentary receptions

We launched the Stats Behind the Stories report and raised awareness of key facts and figures about rare diseases at the Rare Disease Day Parliamentary receptions which took place during February and March 2024. We were joined by 188 attendees including speaker Andrew Stephenson MP, Minister for Health and Secondary Care at the Westminster Rare Disease Day reception, 94 attendees including speaker Jenni Minto MSP, Minister for Public Health and Women's Health at our Scottish Rare Disease Day reception, and over 80 attendees including speaker Professor Iolo Doull, Medical Director at Welsh Health Specialised Services Committee at our Welsh Rare Disease Day reception. Each event included a passionate speaker from the rare disease community who discussed their personal experience of how living with a rare condition has affected them and their family.

We also hosted a virtual UK Rare Disease Day 2024 Four Nations reception with 113 people registered to attend. Speakers from England, Scotland, Wales and Northern Ireland shared learning from the second year of the UK Rare Diseases Framework action plans and a deeper dive into their approaches to improving the lives of people affected by rare conditions.

Raising awareness among healthcare professionals and the public

Through regular workshops with our member charities and support groups and outreach at external virtual events, we grew a consortium of organisations working together to deliver a high impact communications campaign throughout February.

A wide range of charities and support groups downloaded our messages, graphics and materials to use on Rare Disease Day 2024 including: Behcets UK, CJD Support Network, CMTUK, Glut1 Deficiency UK, Lily Foundation, Metabolic Support UK, MyAware, Myotonic Dystrophy Support Group, Nerve Tumours UK, PIP-UK Poland Support Charity, PKD Charity, Ring20 Research Support, the Smith-Magenis Syndrome Foundation UK, TOFS UK, and Vasculitis UK.

We also worked closely with NHS England, the NHS Genomic Medicines Services, Genomics England and our member charity Medics for Rare Diseases so they could raise awareness of Rare Disease Day and key facts and figures about rare diseases with NHS staff and healthcare professionals through their own social media channels and newsletters.

Alongside EURORDIS we developed and launched a school toolkit to raise awareness of rare conditions among 8-12 year olds.

A strong legacy for future advocacy

In addition to underpinning awareness-raising on Rare Disease Day 2024, the central source of data about rare diseases in the UK published this year on our new website provides a strong legacy that we can build on in the coming years. Access to credible, authoritative and trusted data will help Genetic Alliance UK, our member organisations, and the wider rare disease community to advocate more effectively for people living with rare diseases in future.

Rare Resources guides

In 2022/23 we disseminated hard copies of Rare Resources Scotland and Rare Resources Cymru to key health and education professionals across Scotland and Wales. These are information guides for families who have recently received a diagnosis of a genetic or rare condition, are on the journey to receive a diagnosis or have been told their child's condition is so rare they may not get a diagnosis. The resources were developed and produced by Genetic Alliance UK in collaboration with families and support organisations, and provide links to reliable sources of information and support and 'top tips' from families. The resources promote signposting of people with rare, genetic and undiagnosed conditions to specialist charities and support groups.

During 2023/24 we made the Rare Resources Scotland and the bi-lingual Rare Resources Cymru available in an electronic format on our new website so they are easier for families to find and use.

In 2023/24 we also began work to develop a complementary set of Rare Resources England so that we can broaden our signposting and support for people living across the UK. The Rare Resource guides for England are being developed thanks to support from our industry partners, the Sir James Roll Trust, the Hospital Saturday Fund and The James Tudor Foundation.

3. Providing a strong voice for people living with genetic, rare and undiagnosed conditions

A 'Manifesto for rare diseases' ahead of the coming General Election

During March 2024, a 'Manifesto for rare diseases' was shared with all of the main political parties thanks to partnership working between Genetic Alliance UK, the largest alliance of organisations supporting people with genetic, rare and undiagnosed conditions in the UK and the Specialised Healthcare Alliance, a coalition of patient groups and corporate supporters who campaign on behalf of people with rare and complex conditions.

Together, we called on the next government to commit to building on the current UK Rare Diseases Framework by setting new ambitions for improving the lives of people with rare conditions over the next five years. The manifesto sets out our calls to action, including policy changes that could:

- Help patients get a timely diagnosis
- Increase awareness among healthcare professionals
- Improve the coordination of care
- Increase access to specialised care, treatment and drugs.

The manifesto was shared with health ministers and shadow ministers from the Conservatives, Labour, the Liberal Democrats, Plaid Cymru and the Scottish National Party. Together, we asked all of the political parties to consider the UK's genetic, rare and undiagnosed condition population in developing proposals for their manifestos ahead of the coming General Election.

Raising awareness through the media of issues important to our members

During 2023/24 we've raised awareness of the challenges facing people with genetic, rare and undiagnosed conditions through the national media. We've talked about newborn heel prick testing in The Sunday Times, about waiting times for genomic test results in The Times, and the challenges facing families living with undiagnosed genetic conditions on BBC Breakfast News. Most recently our Chief Executive, Louise Fish, took part in a round-table discussion about how we can transform the lives of people living with rare diseases hosted by the New Statesman.

Helping our members to engage with elected representatives across the UK

Genetic Alliance UK are the secretariat for the All Party Parliamentary Group and Cross Party Groups for rare, genetic and undiagnosed conditions in Westminster, the Senedd and Holyrood.

As the secretariat to the All Party Parliamentary Group on Rare, Genetic and Undiagnosed Conditions in Westminster, we have worked to engage parliamentarians on key issues. In response to calls from the community and recent announcements in the rare condition space, the group has hosted events on care coordination, alert cards, cell and gene therapies and research into rare conditions. Over 20 of our member organisations have joined us in-person in Westminster to raise these challenges with parliamentarians, with many more joining these hybrid meetings online.

In Scotland, the Cross Party Group has been active in supporting work to develop a newborn screening pilot research programme for Spinal Muscular Atrophy and the broader issue of newborn screening for rare conditions. The group has looked at how decisions on newborn screening are made in Scotland and will continue to explore this issue in the coming year.

In Wales, the Cross Party Group co-hosted parliamentary drop-ins in the Senedd focused on genomics and raising awareness of newborn screening. The Welsh drop-in sessions enabled us to engage with a third of Senedd Members from across the political spectrum at each event.

Ensuring patient voices are heard by decision-makers

Our goal is to make sure that whenever decisions are made that impact on people living with genetic, rare and undiagnosed conditions, someone representing our community is present to make sure the decision is informed by the views of people with lived experience of these conditions. We prepare consultation responses on behalf of our community, attend meetings of committees and working groups, and disseminate representation opportunities to our members and people living with genetic, rare and undiagnosed conditions.

In 2023/24 we responded to a variety of consultations from the governments across the UK, healthcare and fertility regulators, healthcare delivery partners and other bodies, ensuring that the voices of our members and the rare community are involved in any decision making or policy development relevant to our community. Over the past year we responded to a total of 16 National Institute of Health and Care Excellence (NICE) scoping consultations, submitted patient perspective statements to the Human Fertilisation and Embryology Authority (HFEA) as evidence for 49 preimplantation genetic testing licence requests, and responded to a further 22 consultations by UK governments and the NHS on a variety of topics.

Shared learning roundtable for members on Managed Access Agreements

When NICE recommends a treatment with managed access, NHS England and the company responsible for the treatment develop a Managed Access Agreement (MAA). This is a short-term agreement setting out how people can access NHS-funded treatment so data can be collected to decide whether it should be available on the NHS in the longer term. In response to a member organisation asking for support to contribute to their first MAA, Genetic Alliance UK called together a group of its members with experience of MAAs for a shared learning roundtable to share their expertise with one another. The roundtable highlighted that many support groups faced similar challenges and experiences. We collated these themes into a Managed Access Agreements report and presented them to NICE and NHS England. This has prompted enhanced support from NICE for small charities contributing the views of people with rare conditions to MAAs.

4. Ensure scientific breakthroughs in genomics drive better research and services

Making sure individuals and families benefit from genomic breakthroughs

Science is making great strides forward in genomics. However, people affected by genetic conditions and their families will only benefit if the UK also invests in clinical research programmes and NHS clinical genetics services. During 2023/24 we used high-profile speaking opportunities for our senior team at the Festival of Genomics and the NHS Genomics Healthcare Summit to raise awareness of the need to ensure individuals and families benefit from genomic breakthroughs. It is vital that findings from research projects and day-to-day clinical practice are shared with individuals and families in a timely way to help them make informed decisions about life-long treatment and care.

Supporting the development of pilot SWAN clinics

SWAN stands for syndromes without a name. It is not a diagnosis in itself, but a term used when a child or adult is believed to have a genetic condition and testing has failed to identify its genetic cause. People affected by a syndrome without a name can have a range of different symptoms and each person is likely to be affected differently. However, many children and adults affected by a syndrome without a name have global developmental delay, learning disabilities, physical disabilities or complex medical needs.

During 2023/24 we collaborated with the Welsh Government and Rare Disease Implementation Group to secure ongoing funding for the UK's first pilot Syndromes Without a Name clinic which is based in Cardiff. We coordinated input from the SWAN Cymru Council made up of parents of children affected by undiagnosed genetic conditions to develop Patient Reported Outcome Measures (PROMs) and Patient Reported Experience Measures (PREMs) to measure benefits to patients referred to the clinic.

We also encouraged NHS England to set up two pilot SWAN clinics for adults and children in England. The development of a service specification for England began in 2023/24 with a view to NHS England commissioning two pilot SWAN clinics during 2024/25.

SWAN UK

Genetic Alliance UK is proud to run SWAN UK, 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. The aim of SWAN UK is that every family gets the support that they need, regardless of whether or not they have a diagnosis.

During 2023/24 we welcomed 205 new individuals to our SWAN UK community making a total of almost 4,000 people. We organised 10 online coffee meetups to introduce families who are new to the SWAN UK community to other families with similar experiences. We organised 20 SWAN UK community events across England, Scotland and Wales, to bring members of the community together for activities including visits to theme parks, soft play, farms, theatres and picnic trips to the park.

In November 2023 we were delighted to announce that the National Lottery Community Fund will be helping us to deliver work in England over the next three years. We currently support over 2,470 families in England who have a child or children with an undiagnosed genetic condition, but we know there are far more families that need support. This funding will help us reach more families of undiagnosed children sooner, increase the diversity of our SWAN community, and create more opportunities for family events, advocacy and volunteering.

Undiagnosed Children's Day 2024

Undiagnosed Children's Day is our annual awareness day held on the last Friday in April. It's our chance to make as much noise as we can, increase awareness of undiagnosed genetic conditions and raise funds to ensure SWAN UK can carry on providing support to families who are affected by them.

In 2023/24 Undiagnosed Children's Day took place on Friday 26 April.

Working in partnership with the Wales Gene Park

In Wales, Genetic Alliance UK works in partnership with the Wales Gene Park, an organisation embedded within the infrastructure for genomic research, education and wider public engagement. Working collaboratively, we raise awareness of the benefits of genomics through delivering events such as bi-monthly genomics cafes for the public and young people in Wales and beyond. We also support the involvement of a Patient and Public Sounding Board, made up of members of the rare disease and cancer genomics community to ensure coproduction is at the centre of the Welsh Genomics Delivery Plan.

Equity of access to cell and gene therapies

On behalf of ATMP Engage, a group where stakeholders collaborate on patient and public engagement, we carried out research to better understand the potential inequalities that could exist when delivering cell and gene therapies in the UK (also known as advanced therapeutic medicinal products or ATMPs). We produced a report on ATMPs and equity of access with recommendations to address these inequalities to ensure that everyone with a rare condition who may benefit from these types of therapies has equal access to them. We also facilitated involvement of people with lived experience in the cutting-edge pan-European ARDAT project, to directly influence the progress being made toward streamlining of cell and gene therapy development and regulation.

5. Promoting opportunities to expand screening in line with international best practice

Supporting research on new approaches to genomic screening

During 2023/24 we worked with Genomics England to inform the development of their new research programme focused on whole genome sequencing for newborn babies, the Generation Study.

The Generation Study is an NHS-embedded research study which will explore the benefits, challenges and practicalities of sequencing and analysing newborns' genomes. The study will sequence and analyse the genomes of 100,000 newborn babies in the UK. It is a hybrid clinic-research study that aims to generate evidence on whether whole genome sequencing can be used to screen newborns for more than 200 rare genetic conditions, and to assess the feasibility of doing this within the NHS.

In 2023/24 we co-chaired Genomics England's new Generation Study working group on communicating results and onward support which gave us an opportunity to ensure those crucial first moments on a rare condition journey are delivered appropriately, with a balanced and informative approach to introducing new parents to the risk that their child may develop a rare condition. We were able to bring the expertise of our member organisations who already receive 'screen positive' messages through the newborn bloodspot 'heelprick' programme to this development process. We also sit on the Ethics Working Group for this significant research study.

In September 2023 an initial list of over 200 genetic conditions that will be screened for by the Generation Study was published. Genomics England joined us for a special Member Briefing and hosted virtual drop-in sessions to talk to our members about how this research study could affect charities supporting people living with rare genetic conditions and the people they support.

Our research staff are part of the independent evaluation team (led by researchers at UCL and Great Ormond Street Hospital for Children NHS Foundation Trust) that was awarded the contract to assess the process and impact of the Generation Study, after competitive tender. This work will begin in 2024/25.

Working collaboratively to raise awareness of the potential to expand current newborn screening programmes in the UK

Some of the appetite for genomic screening from families living with genetic, rare and undiagnosed conditions is because the UK is not extracting all of the potential from its conventional newborn and population screening programmes. For example, the newborn bloodspot 'heelprick' test given to every newborn baby in the UK currently screens for a maximum of nine conditions, but there are more than 20 European countries screening for more than the UK, with most of those screening for 20 conditions or more.

We continue to be part of the Newborn Screening Collaborative chaired by the MPS Society. We are also members of the Blood Spot Task Group of the UK National Screening Committee which works towards fulfilling the UK Rare Disease Framework objective of improving the evidence base to help the committee make rapid and robust decisions about newborn screening for rare diseases.

Raising awareness of international best practice

Our approach to the current newborn screening programme is informed by our understanding of international best practice in this area. We work collaboratively with Eurordis – Rare Diseases Europe and their working group on newborn screening.

We also work with Screen4Care, a European project examining newborn screening using genome sequencing, as well as looking at AI tools to examine health records to identify rare conditions. We are members of their Patient Advisory Board and of their multi-stakeholder forum.

This year and the coming year

Delivering the 2023/24 Business Plan

Our 2023/24 business plan focussed on 'doing what we already do better' to tackle internal challenges facing the charity and put us in a strong position to implement our new five-year strategy once approved. Our focus was on:

- Clarifying our offer to member organisations and delivering it to a high standard.
- Launching a new website to improve understanding of what we do and why it matters.
- Building on our strengths of the policy and research teams and their clear focus on
  - implementing the UK Rare Diseases Framework
  - providing a strong voice for the genetic, rare and undiagnosed community
  - driving better research and services for people living with genetic conditions
  - and promoting opportunities to expand screening in the UK.
- Clarifying and improving our offer to the SWAN UK community.
- Improving our approach to fundraising, income generation and corporate services.

We took urgent steps to address the financial challenges facing Genetic Alliance UK through a change management programme focused on cost savings, income generation and reducing headcount. We made cost savings by decreasing our office and storage space. We diversified our income by implementing a fundraising strategy including new approaches to individual giving, community fundraising, corporate partnerships, and in-memory and legacy gifts. We sadly made three posts at the charity redundant. This work was supported by the second year of a sustainability grant from the Wellcome Trust which helped us to develop our new five-year strategy, build our fundraising capabilities, and modernise our approach to equality, diversity and inclusion.

During 2023/24 we also implemented the recommendations made in our governance review by:

- Recruiting four new trustees to increase the independence and diversity of the Board.
- Introducing a new People and Policies sub-committee, alongside the existing Finance and Governance sub-committee, to share the workload and involve more trustees. The new sub-committee will provide oversight for recruitment of trustees and senior staff, HR, pay and reward, and policies in areas such as data protection, health and safety, and safeguarding.
- Amending the Articles of Association in line with best practice for trustee length of service, membership eligibility criteria, and codes of conduct for trustees and members.

Looking forward to 2024/25

2024/25 is the first year of our new five-year strategy working together to improve the lives of people affected by genetic, rare and undiagnosed conditions.

During 2024/25 our business plan will focus on:

- Continuing to address the significant financial challenges facing the charity by prioritising income generation, making the charity sustainable and rebuilding our reserves.
- Providing a strong, unified voice for our community before and after the UK General Election.
- Continuing our strategic focus on delivering the UK Rare Diseases Framework.
- Providing patient and public voice expertise to ensure new research programmes are shaped by the views of people with lived experience, and promoting a culture in academic and health services of valuing and acting on this input.
- Scoping a new strategic programme of work on screening and securing the funds to deliver it.
- Investing in and making the most of the skills and capacity of our committed staff.

We will continue to take urgent steps to manage the financial challenges facing the charity by:

- Recruiting a new Chief Executive who will complement the skills of our senior team with expertise in business development and income generation, governance, strategic delivery, impact measurement, operational management, business planning and financial management.
- Diversifying income by developing new strategic funding partnerships with research funders and LifeArc and securing a core funding grant from the Department of Health and Social Care.
- Making cost savings by closing our remaining office and storage space and consulting with staff on a major restructure in Q1 2024/25 with a view to ensuring the charity is financially sustainable and can rebuild its reserves.

This work will be supported by the final year of a sustainability grant from the Wellcome Trust to continue building our fundraising capabilities, and modernising our approach to equality, diversity and inclusion.

We have moved three strategic priorities into 2025/26 (the second year of our five-year strategy) so trustees and senior staff can focus on making the charity sustainable. These are:

- Scoping a new strategic programme of work on genomics and building relationships with potential partners and funders for this work.
- Finding partners and funders for a major project to map and signpost people with rare conditions to: charities and support groups for their condition; NHS expertise or centres of excellence for their condition; information about any existing treatment options for their condition; and opportunities to take part in clinical trials of new treatments for their condition.
- Finding a long-term home for the SWAN UK community and supporting change management to ensure that the community are well-supported through this process.

Thank you to our supporters and funders

We raise funds for our vital work with and on behalf of members through individual giving, community fundraising, trusts and foundations, corporate supporters and legacies and in memory giving.

Thank you to all of 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 wish to extend our thanks and gratitude to the families and friends who chose to remember their loved ones through a legacy or in memory donation to support our work during 2023/24.

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

- Alexion
- Amicus
- Anthony Walton Trust
- BioCryst
- Catherine Cookson Charitable Trust
- Chiesi
- The Clothworkers Foundation
- Egetis
- The Hospital Saturday Fund
- Inthallo
- Ipsen
- The James Tudor Foundation
- Janssen
- Kyowa Kirin
- LifeArc
- National Lottery Community Fund
- Novartis
- Orchard Therapeutics
- Pfizer
- PTC
- Robert Luff Foundation
- Roche
- Sir James Roll Trust
- St Andrews Charity Fashion Show
- Takeda
- UCB
- Ultragenyx
- Vertex
- Wellcome Trust

Financial overview 2023/2024

Members can obtain full copies of the Annual Report and Accounts 2023/2024 by emailing the Chair at chair@geneticalliance.org.uk. They are also available on the Genetic Alliance UK website, Companies House and from the Charity Commission.