ACCESS TO MEDICINES BRIEFING

All Party Parliamentary Group on Rare, Genetic and Undiagnosed Conditions

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ALL PARTY PARLIAMENTARY GROUP ON RARE, GENETIC AND UNDIAGNOSED CONDITIONS

The key aims of the All Party Parliamentary Group (APPG) are to increase awareness of rare, genetic and undiagnosed conditions in parliament and help to ensure that patients and families affected by these conditions have access to appropriate care and support.

At the inaugural meeting of the APPG in February 2016, the group agreed to:

- Examine the processes in place for accessing rare disease medicines in England, to consider if these processes are working and if they can be improved for the benefit of patients.
- Hold an APPG hearing to hear directly from stakeholders working in the field, including clinicians, representatives from patient organisations, and industry.

Briefing by Rare Disease UK
September 2016
Photo credit: Josh Tucker/Rare Disease UK

RARE DISEASE UK

Rare Disease UK is a multi-stakeholder campaign run by Genetic Alliance UK, working with the rare disease community and the UK's health departments to effectively implement the UK Strategy for Rare Diseases.

ACCESS TO MEDICINES HEARING

Date: 11 October 2016

Time: 8:30am - 10:30am

Venue: Churchill Room, Houses of Parliament

AGENDA

8:30am	Teas, coffees and light breakfast
8:40am	Welcome from Ben Howlett MP, Chair
8:45am	Case study: ataluren – Janis Clayton, PTC Therapeutics – Nic Bungay, Muscular Dystrophy UK – Dr Adnan Manzur, Great Ormond Street Hospital
9:15am	Discussion
9:30am	Case study: everolimus – Jayne Spink, Tuberous Sclerosis Association – Dominic Wake, Novartis Pharmaceuticals – Dr Chris Kingswood, Brighton & Sussex University Hospitals Trust
10:00am	Discussion
10:30am	Close

Rare disease medicines

Many rare diseases are severe and life-limiting. For most individuals or families affected by rare diseases, the day-to-day challenges of managing a severe condition are made worse by the absence of an effective treatment or cure. For some rare conditions, however, new medicines have been researched, developed and licensed by the European Medicines Agency (EMA). Before these can be accessed on the NHS, they have to be evaluated for cost-effectiveness and prioritised for commissioning.

The current system

There are currently eight access routes through which licensed medicines for rare conditions can be evaluated and/or commissioned to enable publicly-funded patient access, managed by either NICE or NHS England. NICE is currently developing a ninth access route, the Abbreviated Technology Appraisal (ATA) process.

1. Single Technology Appraisal (STA) – NICE
2. Multiple Technology Appraisal (MTA) – NICE
3. Highly Specialised Technology (HST) Evaluation Programme – NICE
4. Cancer Drugs Fund (CDF) – NICE
5. Individual Funding Requests (IFRs) – NHS England
6. Commissioning through Evaluation (CtE) – NHS England
7. Specialised commissioning based on a recommendation by a Clinical Reference Group (CRG) – NHS England
8. Clinically Critically Urgent (CCU) funding request – NHS England

There are a number of issues with the current evaluation processes, preventing many rare disease patients from accessing new rare disease medicines, as identified by patients themselves (Genetic Alliance UK, April 2014; Genetic Alliance UK, October 2014). The main routes for evaluating rare disease medicines are detailed below, with a brief discussion of related access problems.

NICE

When a medicine is approved by NICE, NHS England is legally mandated to fund it and make it available to patients within three months. Historically, NICE has not been the primary route through which rare disease medicines have been considered for NHS-wide commissioning. NICE appraised its first orphan medicine in 2009 and since then has evaluated around three per year. An orphan medicine is one intended for the treatment of a condition with prevalence in the EU of not more than 5 in 10,000 (European Medicines Agency, 2016). This accounts for less than 20% of newly licensed medicines appraised by NICE for non-cancer indications. The Single Technology Appraisal (STA) and Multiple Technology Appraisal (MTA) routes are designed as assessment pathways for less rare conditions, and those with existing treatments, and as such rare disease medicines are less likely to be appraised via these routes.

Highly Specialised Technology evaluation programme

The HST evaluation programme is designed for evaluating medicines for small patient populations; however it currently lacks the capacity to effectively appraise all the new rare disease medicines that the EMA will license (approximately 13 each year), as its terms of reference indicate a target of three evaluations a year. Patients believe that the number of HST evaluations carried out should be determined exclusively by the number that meets the revised selection criteria, which includes a consideration of appropriateness (Genetic Alliance UK, April 2014).

The Cancer Drugs Fund

This was introduced in 2010 as a means of enabling patients to access cancer medicines that would otherwise not be funded by the NHS. It has a ring-fenced budget which was initially set at £200 million a year but has since then increased to £340 million. One justification for the CDF is that without the fund some patients would not have access to any treatment for their condition. However, unmet medical need is not a problem faced by cancer patients alone, and many other patients, including those affected by rare conditions, are currently unable to access medicines or therapies for their condition (Genetic Alliance UK, February 2016).

Until July 2016, the evaluation process was managed by NHS England. However following a series of consultations, the CDF was converted to a managed access fund, and the management of the evaluation process fell to the responsibility of NICE (British Medical Journal, February 2016).

NHS England

The remit of NHS England in terms of determining access to rare disease medicines is significant. Since NICE evaluates so few medicines for rare diseases, NHS England is by default the primary appraisal body and commissioner of rare disease medicines. NHS England also has to make decisions, i.e. come up with policies, for all rare disease treatments that NICE does not evaluate.

Specialised Commissioning by NHS England

In 2014, Genetic Alliance UK undertook research to find out the patient perspective on NHS England's commissioning for rare disease medicines (Genetic Alliance UK, October 2014). The Patient Charter that we produced from this work revealed patients' concerns that they are being prevented from accessing the medicines they need because NHS England is poorly organised, overburdened and under-resourced.

Patient groups agreed there are too many steps in the process for evaluating medicines and that NHS England should streamline their unwieldy governance structure so they can make fast and fair decisions. This will ensure their limited finances are spent where they are needed most. NHS England should be clear who is making the decisions about which medicines are evaluated and by whom during topic selection at NICE, and which are funded, and what criteria are used to make these decisions.

Patient groups felt that NHS England's Clinical Reference Groups were under-resourced and as a result were not able to give expert advice, enable consistent decision-making and engage stakeholders effectively. This finding has been acted upon with NHS England's recent CRG review, which is currently being implemented. NHS England's appraisal process needs drastic streamlining and rationalisation to enable timely, patient-focused transparent commissioning of rare disease medicines. Patients also highlighted the importance of the patient voice in deciding which treatments and services should be commissioned by NHS England.

Individual Funding Requests

The IFR process allows patients who would benefit from a medicine to access it even if they fall outside of current commissioning arrangements, providing that making it available is cost-effective. This route is currently used much more regularly than intended because there is a backlog of treatments waiting to be evaluated by NICE and NHS England, meaning that more appropriate access routes are taking longer than they should. As part of the changes made to the CDF earlier this year, IFRs relating to cancer will no longer be considered via the CDF process, and instead will be considered using the single IFR process (NHS England, 2016).

Clinically Critically Urgent

The standard operating procedure for funding requests for Clinically Critically Urgent treatments was published last year after years of misuse of the Individual Funding Request route for this purpose. This applies where there is no NICE Technology Appraisal for the treatment and indication and no NHS England clinical commissioning policy or agreed interim commissioning position, an IFR is not appropriate because the patient is part of a cohort of patients with the same condition at the same stage of the disease, and the treatment is clinically critically urgent because the patient is at risk of imminent significant and irreversible clinical deterioration (life threatening or major loss of function).

Commissioning through Evaluation

Commissioning through Evaluation (CtE) was developed by NHS England as a new way of commissioning specialised services or treatments for which there is currently insufficient evidence of clinical and/or cost-effectiveness to warrant routine commissioning. The programme's budget is essentially used to fund pilot schemes. During the pilot, data is collected which can then be used to support a decision as to whether a specialised commissioning proposal should be funded or not. So far, no medicine for a rare condition has been selected for the CtE programme.

What are the problems?

Methodology

Methods of decision making regarding rare disease medicines have changed multiple times, both with NHS England's processes and the HST process, since they were designed following the Health and Social Care Act 2012.

Patient groups found the original processes for both HST and NHS England's processes to be largely fit for purpose, barring other concerns around communication and transparency described elsewhere in this briefing. However, these methods have evolved significantly since those examinations. Observation of HST meetings have shown that questions outside of the scope of the programme's terms of reference are relatively common, and many of the evaluations have used quality-adjusted life-year (QALY) methodology, which is not part of the original proposed method. We now have the new addition of managed access agreements (MAA), a major new development which already appears to be likely to be part of most future HST evaluations.

There have been two MAAs following HST evaluations to date. These are agreements between the patient community, NHS England, clinicians, and the pharmaceutical company producing the medicine which govern the terms under which the performance of the medicine, and therefore its future commissioning, over a set period. These have come about without a set methodology for development, and the two that have been developed so far are very different (which is not necessarily a negative). They also happen to be for treatments for patient populations which have a well-organised patient support group, which is unlikely to be the case for all future treatments.

NHS England has changed its prioritisation methodology a number of times, sometimes altering one phase in isolation from other components of the process. Issues with transparency pose challenges to assessing the value of these changes. Other method changes have been implemented without consultation or following a poorly coordinated consultation, one which clashed with related policy initiatives such as the Accelerated Access Review, or in some cases a consultation has been expected for several years with as yet no indication when it will take place.

Topic selection

The various routes for access decisions each have their benefits and disadvantages. The selection of a particular process for a particular treatment may be one of the biggest factors that ultimately influences access. It is important that the most appropriate assessment route for each medicine is selected by a process that is both transparent and neutral.

Currently there is no overarching assignment that applies to all assessment routes. NICE runs a process that decides which medicines will be evaluated in its streams: topic selection. Due to a lack of clarity and transparency on this process, information on how or why access routes are selected in topic selection is not available. Those medicines not selected by NICE may be selected by NHS England for one of their access routes, but here there is no organisation-wide approach, and therefore no transparency in decision-making for the selection routes.

The current system creates a significant risk that inconsistent and inequitable decisions are made in assigning access routes to treatments. Without a coordinated, transparent and publicly accountable approach for triaging medicines into each route, there is a risk that some medicines will be not be assigned an access route. Patients requiring access to this treatment are then forced to rely on attempting to gain access on a case-by-case basis through the IFR process.

Communication with stakeholders

NHS England and NICE have a significant number of stakeholders ranging from large, professional organisations to individual patients and the wider public. The information and insight they can provide is valuable at all levels of decision making: from the appropriateness of specific commissioning policies to cross-cutting structural or procedural changes, such as the development of CPAG's funding prioritisation framework or the HST evaluation methodology. Many stakeholders, including patients and patient groups, can also provide feedback on how policies or processes are working on the ground.

Good stakeholder engagement requires information to flow effectively between NHS England and NICE and their stakeholders. This requires both bodies to both provide information to and actively seek out the views of their stakeholders.

Transparency

Patient groups are frustrated about the lack of transparency with the constantly changing processes at both NICE and NHS England. For example, very little information has been provided on how exactly the Clinical Priorities Advisory Group (CPAG) reaches their decisions on which policies fall into each prioritisation category, other than that it will be through debate. The lack of transparency makes it impossible for stakeholders to assess what evidence has contributed to the decision, and whether this has included patient voice.

This lack of transparency is exacerbated by the reluctance of senior NHS England staff to reassure stakeholders that detailed minutes of CPAG's deliberations would be published. Without detailed, publicly available narrative minutes, it will be impossible for stakeholders to tease out the precise reasons why each policy was given its relative position compared to each competing policy. While we understand that the processes are being proposed for 2015/2016 only, and will not necessarily be reused in subsequent years, in order for NHS England to learn from the experience external stakeholders must be able to understand how these decisions are being made and hold decision makers to account.

Conclusions

Addressing the current failings in topic selection is an essential part of ensuring that the system through which patients with rare diseases can access the most cost-effective medicines is fit for purpose. Patient representatives agree that achieving this will require NHS England and NICE to collaborate and deliver a transparent and rational methodology for triaging medicines into the most appropriate access route (Genetic Alliance UK, April 2014). In addition, patient representatives also agreed that, as valuable stakeholders, patients should be given the opportunity to comment on the proposed work streams of NICE and NHS England for their views to be considered before the work streams are finalised.

Without a coordinated, transparent and publicly accountable approach for triaging medicines into each route, inconsistent decisions are at risk of being made, leading to inequitable patient access to medicines. Addressing the current failings in topic selection is an essential part of ensuring that the system through which patients with rare diseases can access cost-effective medicines is fit for purpose. Patient representatives have agreed (Genetic Alliance UK, Oct 2014) that achieving this will require NHS England and NICE to collaborate and deliver a transparent and rational methodology for triaging medicines into the most appropriate access route. In addition, patient representatives agreed that, as valuable stakeholders, patients should be given the opportunity to comment on the proposed work-streams of NICE and NHS England and for their views to be considered before the work-streams are finalised.

The system described above relates specifically to England. The picture for rare disease patients looking to access a medicine for their condition is different if they live in Scotland, Wales or Northern Ireland. In the last few years both Wales and Scotland have developed and implemented a new process for appraising orphan and ultra-orphan medicines. Many of the changes have been welcomed by the rare disease patient community. Working together and sharing best practice across the four nations will be essential for ensuring that all rare disease patients in the UK are able to access the best medicines for their conditions.

Ensuring that all the different routes work effectively together within England is a considerable challenge. Further ensuring that all patients within the UK, regardless of where they live, can access the best treatments and services for their condition will require even greater collaboration between the NHS and health departments of the four home nations. A robust and transparent system in England would form a crucial part of this process.

ATALUREN FOR NONSENSE MUTATION DUCHENNE MUSCULAR DYSTROPHY

Case study one

Duchenne muscular dystrophy (DMD) is the most common and most rapidly progressing form of muscular dystrophy. Affected young people experience progressive muscle weakness due to a lack of the protein dystrophin. DMD is typically diagnosed in early childhood when the affected individuals have difficulty walking, standing, or climbing stairs, and may have behavioural or learning difficulties. After the age of 12 most children will need to use a wheelchair. During adolescence, breathing muscles can weaken, causing shallow breathing and a less effective cough mechanism, which can lead to chest infections. Weakness of the heart muscle, called cardiomyopathy, occurs in almost all patients by the age 18.

The life expectancy of people with DMD depends on how quickly and intensely muscle weakness progresses and on how it affects the patient's ability to breathe. The average lifespan is less than 30 years. Corticosteroids are currently used to help manage the symptoms of the disease; however they do not treat the underlying cause. Nonsense mutation Duchenne muscular dystrophy (nmDMD) is one of several genetic variants of DMD, affecting around 10% of children and young adults living with DMD in England.

The medicine ataluren (Translarna, PTC Therapeutics) was granted marketing authorisation in Europe in July 2014 and is the first approved medicine that addresses the underlying cause (in affected individuals with amenable mutations) by enabling the body to produce its own dystrophin.

As ataluren is a medicine with a novel mode of action intended to treat a subpopulation of a rare condition, the NICE Highly Specialised Technology (HST) evaluation process – designed specifically to evaluate medicines for small populations – would appear to be the most appropriate appraisal route. However, the published interim prioritisation criteria state that to be eligible for evaluation through this route, the affected patient population must be 'clinically distinct'. Since current interpretation of the term 'clinically distinct' by NICE does not recognise subgroups of patients within patient populations defined by genetic or biomarker differences only and where the clinical presentation itself does not markedly differ, a medicine to treat nmDMD did not meet the criteria for an HST evaluation.

From July 2014 the Paediatric Neuroscience clinical reference group (CRG) began working on a commissioning policy for ataluren, with the assistance of PTC Therapeutics. Following the CRG's approval of the policy, the evaluation progressed to the Women and Children's Programme of Care Board who also approved it. In October 2014, the evaluation then went to the Clinical Priorities Advisory Group (CPAG) prioritisation meeting. CPAG members raised a number of issues to be addressed by PTC Therapeutics. The months that followed involved a number of further meetings and discussions between the CRG and PTC Therapeutics with the expectation that the ataluren policy would be reviewed at the December 2014 CPAG meeting.

In December 2014, legal challenges to the validity of the CPAG prioritisation process forced NHS England to reconsider how it assesses new treatments through the specialised commissioning route. In the midst of a significant build-up of political activity surrounding this, the scheduled CPAG meeting considering whether to fund ataluren for nmDMD was postponed while NHS England carried out a 90-day consultation on prioritisation for specialised services.

In January 2015, DMD patient support groups engaged in substantial lobbying and campaigning work, expressing their frustrations about ongoing delays and uncertainty. Shortly afterward, George Freeman MP requested that ataluren be reviewed by NICE, despite it not meeting the HST selection criteria. In March 2015, NICE held an initial scoping meeting to commence the evaluation process through the HST access route. The following month, at the NICE decision problem meeting, PTC Therapeutics was informed that they would need to submit the evidence review by mid-June in order for the drug to progress through the stages of the HST evaluation.

In parallel with these timings, and following the prioritisation consultation process, the CPAG process was re-started. Discussions continued regarding the CRG policy for ataluren, however, following the June 2015 CPAG meeting it was announced that NHS England could no longer consider ataluren as it was now on the NICE work programme. Until this point, the manufacturer, patient groups and clinicians had continued to engage with the commissioning policy process while the NICE process was taking place, believing that this might enable an interim funding policy.

In October 2015, the HST Evaluation Committee published a negative interim decision pending additional phase 3 trial results and further questions to be answered with regard to value. PTC Therapeutics submitted further evidence and a second NICE HST committee meeting was held in November 2015. During this meeting, NICE stated that they required a managed access agreement (MAA)to be developed.

During December and January discussions took place between PTC, NHS England, physicians, patient organisation representatives and NICE to develop the MAA. The clinical aspects of the MAA were agreed by this stakeholder group. The process also required that a financial negotiation take place between NHS England and PTC Therapeutics. These financial discussions took place in mid-February in order that the outcome of the MAA discussions (clinical and financial) could be submitted to NICE in time for their late February committee meeting.

In April 2016, NICE published a positive recommendation for ataluren. However, in a press release that was also published on the same day, it was indicated by NICE that PTC Therapeutics and NHS England would need to further negotiate the terms of the commissioning. In May 2016, NICE issued a further press release stating that they were extending the appeal period following the positive recommendation, placing NHS England and PTC Therapeutics under eight weeks' notice to reach an agreement. If an agreement was not reached NICE would state that the guidance could not be implemented. It was not made clear why the appeal period had been extended, especially given that there had been no appeals, nor why an eight week time frame had been proposed. PTC Therapeutics and NHS England met and agreed a plan of action for implementation, which was ratified by NICE in July 2016.

EVEROLIMUS FOR SUBEPENDYMAL GIANT CELL ASTROCYTOMA IN TUBEROUS SCLEROSIS COMPLEX

Case study two

Tuberous Sclerosis Complex (TSC) is a rare genetic condition estimated to affect one million people worldwide. The condition results in the development of non-cancerous tumours, often in the brain, eyes, heart, kidney, skin and lungs. The size and location of these growths determines the impact of TSC, with some people being relatively mildly affected and others more severely. Symptoms may present early in life or not until adulthood.

In September 2011 the European Medicines Agency (EMA) approved everolimus (Vortubia, Novartis) for use in Europe as the first medicine licensed to treat a type of brain tumour (subependymal giant cell astrocytoma or SEGA) in children and adult TSC patients where surgical removal is not an option. The following year everolimus was also granted a licence for treating a type of kidney tumour (renal angiomyolipoma or AML) in adult TSC patients who are at risk of complications but do not require immediate surgery.

For the first twelve months after licensing, there was no routine commissioning policy for everolimus for SEGAs. At that time patient access was possible only through individual funding requests (IFRs), either to Primary Care Trusts or to the Cancer Drugs Fund. Although applications to the CDF were generally more successful than applications made to PCTs, most funding applications were rejected because they were not sufficiently unique to satisfy the exceptionality criterion.

Following the introduction of the Health and Social Care Act 2012, NHS England became the body most likely to be responsible for evaluating everolimus. In February 2013 the Tuberous Sclerosis Association (TSA) met with NHS England and provided an evidence pack and business case on everolimus which they had prepared.

In April 2013, NHS England's Multi-System Disorder Clinical Reference Group (CRG) began work on a commissioning policy for everolimus for both licensed indications, as a well as a service specification for treating those affected by TSC in England. However in December 2014, this work was suspended and the CRG was formally disbanded in February 2015. As the policy had not yet been completed or approved, no funding was available for everolimus.

A Freedom of Information request lodged by the TSA in mid 2015 revealed that around 30 IFRs had been declined by NHS England, as the number of patients requesting the medicine via an IFR had reached the threshold to trigger a cut-off in access via the IFR system and simulate the development of prescribing policies by NHS England.

Following a consultation on a new prioritisation framework in January 2015, the process of developing commissioning policies for everolimus was restarted in July 2015: in the Paediatric Neurosciences CRG for SEGAs, and the Specialist Urology CRG for AMLs.

In June 2015, following much campaigning by the TSA, significant media and political interest and a letter from 20 NHS consultants to the Board of NHS England highlighting the situation, NHS England published the clinically critically urgent standard operating procedure policy for access to medicines. Also in June 2015, an NHS England Specialised Services Circular committed to the development of in-year policies for TSC related brain and kidney tumour treatment. Such policies would allow access for those with advancing disease who meet strict prescribing criteria, but who are not at imminent risk of death.

The draft policy for everolimus for inoperable SEGAs was finalised by the paediatric neurosciences CRG, sent to stakeholders for review and a public consultation held. This process was completed in good time for it to be published in-year (2015/16) and NHS England agreed that the evidence was in favour of routine prescribing. Despite this, the policy was not considered for in-year commissioning.

The everolimus for SEGA policy was then included with a significant backlog of policies sent to the June 2016 meeting of the Clinical Priorities Advisory Group (CPAG) to be considered for funding for 2016/2017. The policies were subjected to a new prioritisation mechanism which significantly disadvantages medicines for rare diseases. When the provisional outcomes of the CPAG prioritisation process were announced in July 2016, everolimus for inoperable SEGAs was listed as having been ranked in priority level 5, meaning that in the opinion of the members of CPAG the policy was of the highest cost and lowest benefit groupings.

The outcomes of the June CPAG meeting were only provisional because at that time NHS England was awaiting the outcome of a judicial review by the National AIDS Trust, challenging NHS England's claim that they did not have the legal power to commission pre-exposure prophylaxis (PrEP), a method of HIV prevention. At the beginning of August the High Court ruled in favour of the National AIDS Trust, and NHS England announced they would be appealing the decision. In the meantime, NHS England has been consulting on a commissioning policy for PrEP, and CPAG will be asked to re-run the prioritisation process as soon as practicable, likely in October 2016. They will assign each policy (PrEP and the policies and treatments from level 3-5 of the previous prioritisation process) to one of five priority levels relative to the other policies considered, using the same process as at the June CPAG meeting.

However, the end of August 2016 NHS England sent a letter to companies manufacturing a treatment which would be included in the October CPAG prioritisation, detailing what they termed a soft market testing process. In this process, companies would be able to see the relative ranking of each commissioning policy based on the cost per patient values. Companies would then be able to adjust the cost per patient for their medicine, and see the ranking change accordingly.

At this point it is not clear what the outcome of this further delay will be for patient access to everolimus for SEGAs due to TSC. Depending on the rankings produced through the soft market testing process and the outcome of the October CPAG prioritisation, we may see a policy or treatment not previously prioritised for commissioning ranked higher against alternative interventions by CPAG, and so be able to be commissioned. If NHS England is successful in its appeal, on the other hand, the prioritisation decisions made in June will stand, and these will be implemented, including a decision not to fund routine commissioning of everolimus for SEGAs due to TSC.

Leadership

Ben Howlett MP, Chair

Before being elected as the Member of Parliament for Bath in May 2015 Ben worked alongside the National Health Service as a senior consultant and has over 8 years experience in the health sector.

Since his election, Ben is an active member of both the Women's and Equalities Select Committee and the Petitions Select Committee.

Vice-Chairs

Rt Hon Cheryl Gillan MP for Chesham and Amersham. During Cheryl's time in Parliament she has served as the Secretary of State for Wales.

Lord Turnberg is a leading member of the APPG for Medical Research. He is a medical professional and was previously the President of the Royal College of Physicians.

Rt Hon Baroness Pauline Neville-Jones DCMG PC has previously served as Minister of State for Security. She is also the patron for the charity Unique – the Rare Chromosome Disorder Support Group.

Lord Patel KT is an obstetrician by profession. Currently Lord Patel is the Chancellor of the University of Dundee and President of the charity Attend.

The secretariat to the APPG on Rare, Genetic and Undiagnosed Conditions is provided by Genetic Alliance UK, the national charity working to improve the lives of patients and families affected by all types of genetic conditions. Genetic Alliance UK an alliance of over 180 patient organisations.

What we do

Supporting: We seek to raise awareness of genetic conditions and improve the quality of services and information available to patients and families.

Campaigning: We actively campaign on behalf of those with genetic conditions on issues of policy and practice to influence governments, policy makers, industry and care providers such as the NHS.

Uniting: We provide a united voice for all those affected by genetic conditions, enabling us to work together towards the common goal of making life better for patients and families at risk. We are the only organisation in the UK that provides a voice for all patients and families affected by genetic conditions on a European, UK and devolved nation level.

Our aim is to ensure that high quality services, information and support are provided to all who need them. We actively support research and innovation across the field of genetic medicine.

Rare Disease UK is a multi-stakeholder campaign run by Genetic Alliance UK, working with the rare disease community and the UK's health departments to effectively implement the UK Strategy for Rare Diseases.

SWAN UK (syndromes without a name) is a patient and family support service run by Genetic Alliance UK. SWAN UK offers support and information to families of children with undiagnosed genetic conditions.