



**Genetic Alliance UK**  
Supporting. Campaigning. Uniting.

## Facilitating Networks of Expertise



## Genetic Alliance UK

Genetic Alliance UK is the national alliance of 150 charities and support groups for children, adults and families who are affected by all types of genetic disorders. We aim to improve the lives of people affected by rare genetic conditions by ensuring that high quality services and information are available to all who need them. Our work includes providing accessible and accurate information about genetics, genetic disorders and genetic issues and working with our member groups and with health professionals to improve access and services for people with genetic disorders. Our also play an advocacy role, representing and campaigning on behalf of people who might otherwise have no voice or influence on government policy or on the provision of the medical and care services on which they depend.

Genetic Alliance UK undertakes various projects and programmes that provide evidence and knowledge on which to base improvements to health service provision, research and support for families. These initiatives include:

- Rare Disease UK, a multi-stakeholder group working to provide information to and persuade health departments and the NHS to develop a plan for the provision of services for patients with rare diseases. The provision of accurate and appropriate information is an essential element of this.
- Syndromes Without A Name (SWAN), a UK-wide network providing information and support to families of children without a diagnosis.
- Route Maps for Rare Conditions, a project through which ten of our small member groups are developing a practical and cost-effective framework for improving information, access and coordination of health and social care services for individuals and families with a wide range of rare genetic conditions.
- Family Route Maps, a pilot project developing a tool to help families with genetic conditions to access appropriate information and services in the UK, worked in partnership with individuals and families affected by a range of genetic conditions.

To download a copy of this document, please visit the following page:

[www.geneticalliance.org.uk/projects/facilitatingnetworks.htm](http://www.geneticalliance.org.uk/projects/facilitatingnetworks.htm)

Published by:	Genetic Alliance UK Unit 4D, Leroy House 436 Essex Road London, N1 3QP
Telephone:	+44 (0)20 7704 3141
Fax:	+44 (0)20 7359 1447
Email:	contactus@geneticalliance.org.uk
Website:	www.geneticalliance.org.uk
Registered Charity Numbers:	1114195 and SC039299
Registered Company Number:	05772999
Cover illustration by:	Rebecca Kent rebeccakent.com
Published date:	December 2011

## Facilitating Networks of Expertise

## Author

Krystle Kontoh – Genetic Alliance UK

## Acknowledgements

### Hypomelanosis of Ito Network

Terri Grant – HITS UK

Dr Saleem M Taibjee MB BCH BMedSci RCPC DipRCPath (Dermatopathology) – Birmingham Children's Hospital

### Ataxia Network

Dr. Julie Greenfield – Ataxia UK

Marios Hadjivassiliou MD – Sheffield Hallamshire Hospital

Dr. Alison Stevenson – Ataxia UK

### Rasopathies Network

Michelle Ellis – The Noonan Syndrome Support Group (UK Representative)

Patricia Hannan – UK CFC Syndrome Representative

Dr Sue Huson – St. Mary's Hospital, Manchester

Dr Bronwyn Kerr – School of Medicine University of Manchester

Professor Michael Patton – St George's Hospital, London

Lindsey Rennard – Neuro Foundation

Colin Stone – International Costello Syndrome Support Group

Cath Stone – International Costello Syndrome Support Group

### FAP Network

Miss Sue Clark MA MB BChir MD FRCS (Gen Surg) – St. Mark's Hospital, London

Peter Granger – PolyPeople

Dr Fiona Laloo – Manchester Children's University Hospitals

Mick Mason – FAP Gene UK

John Roberts – FAP Gene UK

### Cerebral Cavernous Malformations Network

Dr Rustam Al-Shahi Salman – University of Edinburgh

Dr Jonathan Berg – University of Dundee

Mr Neil Kitchen – National Hospital for Neurology and Neurosurgery

Dr Ian Stuart – Cavernoma Alliance UK

### Websites of the patient groups involved

Ataxia UK:

[www.ataxia.org.uk](http://www.ataxia.org.uk)

FAP Gene UK:

[www.fapgene.org.uk](http://www.fapgene.org.uk)

Genetic Alliance UK:

[www.geneticalliance.org.uk](http://www.geneticalliance.org.uk)

HITS UK:

[www.e-fervour.com/hits](http://www.e-fervour.com/hits)

International Costello Syndrome Support Group:

[www.costellokids.com](http://www.costellokids.com)

Neuro Foundation:

[www.nfauk.org](http://www.nfauk.org)

PolyPeople:

[www.polypeople.net](http://www.polypeople.net)

## Contents

Acknowledgements.....	4
Executive summary .....	6
Introduction.....	8
Gathering Evidence.....	9
Surveys .....	9
Focus group summary .....	12
Network formation; principles & practice .....	15
Processes of network development.....	15
Configuring networks.....	16
Redefining the concept of a network.....	16
Network of expertise for Hypomelanosis of Ito.....	18
Network of expertise for Ataxia UK .....	20
Network of expertise for Rasopathies.....	21
Network of expertise for Familial Adenomatous Polyposis .....	23
Network of expertise for Cerebral Cavernous Malformations .....	25
Conclusion .....	26

## Executive summary

### Background

Tackling health inequalities is an integral part of the Westminster government's healthcare reform agenda, to which they have made explicit commitments. Whilst appreciating the complexity of factors determining such inequalities, one might confidently propose that access to appropriate and timely healthcare services and treatments remains a central contributory feature of any successful strategy to overcome those inequalities.

As many patients and families affected by rare genetic diseases often report difficulties in accessing healthcare services and information relating to their condition, there is an increasing awareness of the vulnerability this patient population has to becoming and remaining disenfranchised.

Genetic Alliance UK's mission to support such individuals and families is demonstrated by its history of project work dedicated to understanding the experiences of patients and families affected by rare genetic conditions, and utilising its in-house skills and expertise to work with such groups to improve this situation.

### Project details

The Facilitating Networks project was designed to address the difficulties that children, adults and families with rare genetic disorders experience in obtaining good information and optimal healthcare for their condition. The primary aim of the project was to work with small patient support groups to facilitate the development of networks of health and social care professionals in order to improve information, care and services for patients and families affected by, or at risk of, rare genetic disorders.

The groups selected to participate in the project were small, under resourced, and had no clinical network already in existence. They included:

- HITS representing patients and families affected by Hypo Melanosis of Ito.
- Ataxia UK representing patients and families affected by Ataxia.
- Cavernoma Alliance UK (CA UK) representing patients and families affected by Cerebral Cavernous Malformations.
- FAP Gene UK representing patients and families affected by Familial Adenomatous Polyposis.
- PolyPeople representing patients and families affected by Familial Adenomatous Polyposis.
- International Costello Syndrome Support Group (ICSSG) representing patients and families affected by Costello Syndrome and Cardiofaciocutaneous (CFC) syndrome in the UK.
- Neuro Foundation representing patients and families affected by neurofibromatosis.
- Noonan Syndrome Association representing patients and families affected by Noonan Syndrome in the UK.
- The Noonan Syndrome Support Group (TNSSG) representing patients and families affected by Noonan Syndrome in the USA and the UK.

Five networks were developed across four condition areas, namely a rare dermatological network, a rare cancer network, two networks for rare neurological conditions, and network for Rasopathies.

The aims and objectives for each network were established following extensive consultations with members of their constituent patient support groups. Areas of work focussed on educating healthcare professionals, developing clinical guidelines and enabling expert patients to share their knowledge and experience with other patients.

As a pilot project, this work has evidenced the benefits and implications for support groups and the families they represent working in partnership with clinicians. Genetic Alliance UK anticipate that as a result of the work undertaken there will be a potential improvement in the information, care and services for approximately 50,500 people with rare genetic disorders in the UK.

## Introduction

Currently in the UK, one baby in 33 is born with a genetic disorder or birth defect. When children, adults and families are diagnosed with a genetic disorder, they often experience difficulties accessing satisfactory information, optimal treatment and monitoring of their condition. In many instances this situation is exacerbated when the condition is rare, as neither the family nor the clinician has heard of it before diagnosis.

Whilst such cases illuminate the disjointed nature of patient pathways, some anecdotal accounts also reveal pockets of good practice that may offer insight into how health care professionals can develop appropriate patient-centred models of clinical practice.

When Genetic Alliance UK surveyed its members' experiences of accessing healthcare services, its findings illustrated that clinical networks, where they did exist, made significant improvements to the self-reported health and wellbeing of patients. Even for patient groups where there were no networks in place, patient representatives still identified these as beneficial. The formal and informal networks of clinicians within each condition area appeared to help support the groups in obtaining more knowledge of best practice for their condition, and to create contacts amongst the experts within that field.

"linked groups of health professionals and organisations from primary, secondary and tertiary care, working in a co-ordinated manner, unconstrained by existing professional and health board boundaries, to ensure equitable provision of high quality clinically effective services"

The pattern that emerged from the review of survey responses demonstrated that patient groups representing more commonly recognised conditions, which in most instances had paid staff members, were more likely to have established networks when compared to patient groups representing rarer conditions, which tended to be inadequately resourced.

It was against this background that the Facilitating Networks of Expertise project was conceived. The project's underlying objective was to address, through the development of networks of expertise for groups of rare genetic disorders, the difficulties that children, adults and families with rare genetic disorders experience in receiving good information and optimal healthcare for their condition.

The processes of patient engagement and outcomes achieved through the collaborative efforts of patients and clinicians have been reported so that other patient support groups may also benefit from the lessons learned.

## Gathering evidence

From December 2008 to June 2010 the Facilitating Networks Project undertook a series of patient engagement activities. This process was designed to identify what issues were of primary concern to the patients and families the networks were to represent.

Both quantitative and qualitative methods were used to gather information relating to various aspects of patients' experiences. A summary of findings have been presented below.

### Survey of the Neuro Foundation's members

From the 200 questionnaires sent to members of the Neuro Foundation, 149 were completed and returned to Genetic Alliance UK.

The questions posed sought to measure the extent to which patients and families were satisfied with the health care they received, and to gain insight into their experiences of accessing these services.

**Finding: there was a general lack of support for managing the day-to-day difficulties that are experienced as a result of living with Neurofibromatosis 1 (NF1).**

As many of the local support groups coordinated by members of the Neuro Foundation were meeting less frequently, the patient group was keen to hear where their members were now seeking support to address the psychosocial problems that were known to be associated with the condition.

Overall, many of the respondents described a lack of support for patients with NF1. Individuals stated that they did not receive any support with the ongoing management of their condition either because they felt it was not needed, or they made reference to the dearth of support services in their locality. A common answer was:

**"I don't really have any support from anyone."**

Those who felt they needed occasional support, particularly in relation to social care matters, explained that when necessary they would refer to information provided by the charity's newsletter, educational literature or perhaps contact an NF Specialist Advisor. A few of the respondents referred to the support they received from their GP, specialist consultant and regional genetic service.

**Finding: an overwhelming number of NF1 patients want more information relating to their condition.**

When asked whether individuals felt they would benefit from more information about their condition, 89% stated that they did and specified the particular aspects of their condition they wanted more information about as follows:

- 44% wanted to access information relating to genetic testing.
- 83% wanted more information about medication, treatments and surgical procedures.
- Clinical research updates (75%).
- "Other" information: primarily referring to support services in the education and social care sectors.

Note: these options were not mutually exclusive.

Anecdotal accounts suggested some members of the Neuro Foundation had experienced difficulties understanding medical information communicated by the healthcare professionals they had come into contact with. However, when this question was posed, 76% of respondents stated that they did understand the majority of information given to them. These respondents elaborated, explaining that GPs and nurses seemed to have very limited knowledge about the condition or where to direct them for further information and, in such cases, individuals were able to understand information provided because it was basic and limited. Many of the comments made were akin to:

“My GP does not know much about NF.”

The comments from the 8% of respondents who felt they were not able to comprehend such information seemed to refer to the lack of communication from healthcare professionals rather than an inability to make sense of information shared during the consultation process. Amidst these responses there were a minority that expressed problems when attempting to make sense of medical terminology.

Finding: there is a lack of communication between Healthcare professionals involved in the care management of NF1 patients.

Concerns relating to communication processes were not solely restricted to patient - clinician consultations. 45% of respondents felt there was a lack of communication between the healthcare professionals involved in managing their care.

It would appear that communication breakdown extended beyond individual clinicians, as a number of respondents voiced grievances relating to the transmission of patient information between different hospitals where the patients were receiving care.

“There is a lack of communication between different hospitals and poor communication between specialists.”

Many also noted that receiving care from more than one hospital was time consuming, and there was a lot of travelling involved for those who required care outside of their area.

Finding: patients who had been in the care of a multidisciplinary team reported improvements in the quality of care they received.

67% of respondents who reported having been under the care of a multi-disciplinary team explained their quality of care significantly improved as a result of having access to specialists with expertise in diagnosing and treating NF. Several respondents also commented that this model of healthcare delivery reduced the stress caused by attending multiple appointments at different hospital sites.

Finding: members of the Neuro Foundation supported the idea of a clinical network for their condition.

In order to seek participants' views on the idea of a clinical network for NF, respondents were given the working definition of a network and asked whether they thought such a model would improve their access to specialist healthcare and expertise. Although this was a hypothetical question, 79% responded positively in support of the concept. Many of these respondents commented that the success of the network would be dependent on the communication between healthcare professionals involved.

## Cavernoma Alliance UK questionnaire summary

From the 110 questionnaires sent to members of the Cavernoma Alliance UK, 59 were completed and returned to Genetic Alliance UK.

The questions posed sought to measure the extent to which patients and families were satisfied with the health care they received, and to gain insight into their experiences of accessing these services.

### Information

When asked whether individuals thought they would benefit from more information about their condition:

- 85% responded “yes”.
- 11% responded “no”.

Respondents were given the option to elaborate on what aspects of their condition they would like to learn more about:

- 83% wanted to access information relating to genetic testing.
- 87% wanted more information on medication and treatment options.
- 100% wanting to be kept up to date on clinical research.

Note: these options were not mutually exclusive.

Where individuals were encouraged to elaborate on what ‘other’ information they would find useful, the following responses were made:

“I would have liked to have had some follow-up after my genetic test.”

“Anything available, I have had to find everything myself via the internet.”

“What impact will this have on my dependants?”

### Communication

Individuals were asked in what form they preferred to receive this information.

- 52% opted for paper based text information (e.g. monthly newsletters and journals).
- 52% preferred computer based text information (e.g. using the internet, e-journals).
- 61% preferred to receive information through attending discussion meetings with professionals who are knowledgeable about the condition.
- 30% preferred joining on-line forums and sharing information electronically.

Note: these options were not mutually exclusive.

Individuals were then asked whether they felt there was enough communication between the professionals involved in the care and management of their genetic condition:

- 32% felt that there was.
- 57% stated there was not.
- 11% did not respond.

“What communication! There is no care and management”

“My neurologist did not know much about the condition, which initially exacerbated anxiety”

### Patients' opinions of different models of health care delivery

When respondents were asked whether they had ever been under the care of a multi-disciplinary team (MDT):

- 61% had never been under the care of a MDT.
- 32% had been in the care of a MDT.
- 7% did not respond.

Individuals who had been in the care of a MDT made referred to the benefits they experienced. A few of their comments have been shared below:

“Excellent treatment plan – joined up thinking communication and skill share.”

“Coordinated approach of specialists in different but complimentary fields.”

When given the working definition of a ‘clinical network’ i.e. “linked groups of health professionals from across all sectors of health care e.g. neurology, dermatology who work in a coordinated manner, to ensure equitable provision of high quality and clinically effective services” and asked whether they thought such a network of clinicians with interest in their particular disease group would be a good idea, 93% of respondents were in favour with 7% remaining unsure.

### Focus Group Summary

Focus groups were conducted with members of Ataxia UK, FAP Gene UK and the HITS support group. Although some participants' experiences were related to their particular condition, the central and recurrent themes that emerged were common to each of the groups. These are summarised below.

Core themes:

- Delays in diagnosis and referrals to appropriate services
- Navigating fragmented services
- Accessing information expertise
- Communication

### Delays in diagnosis and referrals to appropriate services

Irrespective of differences in the age of onset and symptoms associated with each condition, many of the participants reported experiencing long delays before receiving an accurate diagnosis for their condition.

“We waited 13 years for my daughter's diagnosis... I think it took so long because [the doctors] didn't actually marry up all the symptoms.”

Patients and parents (in the case of HI) described feeling as though they were continuously being bounced around the healthcare system going from one clinician to another – but remaining unable to obtain the expertise needed. There was also a grave sense of despondency as many patients felt that their concerns had not been taken seriously by the healthcare professionals they saw.

“I was obviously not being listened to, which was very frustrating.”

Participants reported experiencing delays at each critical juncture when being referred to specialist services.

“It's the waiting lists I find most frustrating...You eventually get the referral and then you've got to wait. And then getting the appointments cancelled...the amount of times I've got [my daughter] prepared for an operation and then it's cancelled the day before.”

### Navigating fragmented services

Many participants expressed difficulties when accessing what one patient described as “disparate services” expected to be much more “integrated” in the patient care pathway. These challenges extended beyond the realm of healthcare services as patients described similar frustrations within social services and the education system. Patients recollected the challenges they faced when initially trying to access services, and the poor coordination that ensued.

“You have all these doctors and professionals involved, but it’s you [the patient] holding it all together.”

Participants explained how seeing different specialists for various aspects of their condition had resulted in a multiple diagnoses for individual symptoms and in some instances being initially misdiagnosed.

“I am a whole person but they will only label parts of you.”

There were a number of participants who described positive experiences when a professional adopted the role of coordinating appointments. Two key professionals identified were specialist nurses and social workers.

“My social worker is like a kind of gateway to different services, like direct payments which is fantastic. I mean it’s totally transformed our lives. So if you’ve got a really good social worker that’s really going to help you.”

### Accessing information and expertise

Amongst the myriad of emotions patients reported feeling after receiving an accurate diagnosis, fear relating to uncertainty was often the most prominent. Patients and family members described information seeking as a coping mechanism and reiterated the importance of healthcare professionals providing accurate information relating to the condition at the point of diagnosis. Individuals also insisted on the need to be signposted to reliable sources which they could refer to at a later stage.

Many of the participants used the internet as a primary source of medical information, although at the point of diagnosis several individuals reported feeling overwhelmed by the sheer volume available. There were also concerns about the accuracy of the content provided on the internet.

The greatest challenge many of the patients faced after being diagnosed was finding a specialist for their condition who could manage the coordination of their care over time. Several of the patients recounted the “fight” for access to specialist centres (where they existed).

### Communication

Challenges relating to doctor-patient communication were expressed by the majority of patients who shared their experiences.

Patients commented that they did not feel as though they were being ‘listened to’ by medical professionals, with some mentioning that they believed this had led to the delayed response to their requests for diagnostic tests and/or access to treatments. A number of these participants also described their exasperation when having to “start from scratch” in order to explain their medical history every time they saw a new specialist who had not read patient notes or received adequate handover notes from the referring clinician.

“...it was a sometimes two or three appointments in a week and that’s just a nightmare... and each time I have to explain the same things, and I’m thinking ‘who’s the specialist here?’ It gets really tiring and increases your stress levels thinking the doctor doesn’t know anything about your condition.”

Communication appeared to be undermined in instances where the consulting clinician was unable to provide sufficient information relating to the condition and thus fell short of patient expectations.

“She said ‘I think she’s got HI’ I said ‘what is this?’ and then she said ‘well, go on the net, there is not much to read about it.’”

In some cases patients felt that the lack of information communicated was due to the reluctance of clinicians who were “holding back” information rather than a reflection of their competency.

## Network formation: principles & practice

Having collated information generated by the surveys, focus groups and interviews undertaken, the key issues were identified by the condition group and used to inform the objectives underpinning each of the networks. The processes of network development, activities undertaken and outcomes achieved have been detailed in this section.

### Process of network development

From the outset of this project, the participating patient support groups expressed an interest in the processes of network development, and recognised the value of learning from the successes and failures of patient groups who had undertaken similar work. It was agreed that the project officer would investigate and examine the characteristics of Genetic Alliance UK members who had established networks of expertise for their particular disease area.

Guidance was sought from the following groups:

- The Cystic Fibrosis Trust.
- Primary Ciliary Dyskinesia.
- Alström UK.
- Niemann Pick Disease Support group.

This research enabled the project to observe practical applications of network theory. From these observations, the project team were able to construct a 'checklist' highlighting the fundamental issues participant groups would need to consider at various stages of network development.

- **Network initiation:** each network began with some form of initiation. The common practice, evidenced within each of the groups, was the identification of and invitation to all key stakeholders to contribute to this initial formation.
- **Agreeing the overall purpose of the network:** this involved establishing what areas of work/clinical practice to focus on whilst acknowledging the limitations imposed by finite resources. A mission statement clearly outlining the terms of reference between constituent groups was usually agreed upon.
- **Network evolution:** within the groups consulted there were three distinct ways in which the networks developed; these could be classified as: (i) Patient led (ii) Clinician driven or (iii) Research focussed.
- **Identifying areas of collaboration:** this was largely related to the overall purpose of the network. All of the networks studied focused on one particular area of collaboration (e.g. developing standards of care). Identifying synergies across constituent groups seemed to provide the impetus for a concerted approach that would benefit all stakeholders.
- **Network coordinators:** this described the individuals who adopted the role of intermediaries. These individuals performed the central function of effectively managing inter-organisational and inter-professional relations. In the case of the five networks observed, this took the form of a lead clinician, specialist nurse or a representative from the patient support group. As this individual often served to harness the efforts of all parties concerned, this role was perceived as essential to the continued effectiveness of each network.

## Configuring networks

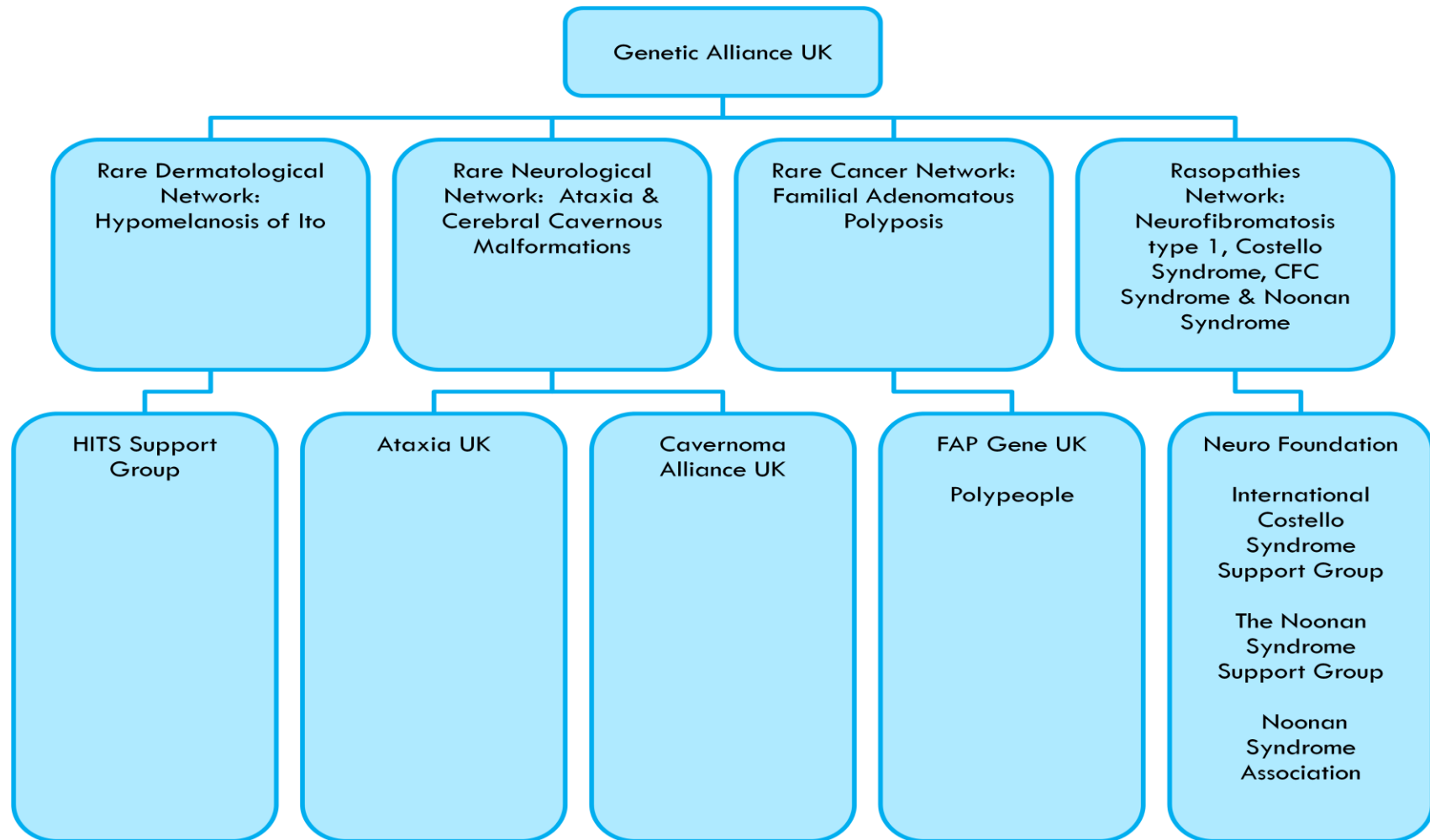
Networks were originally categorised within the three classifications of rare dermatological conditions, rare neurological conditions, and rare cancers. These groupings were later changed as the network activities progressed.

The diagram in table 1 on page 17 represents the final classifications made across each of the participant patient groups and the networks formed.

### Redefining the concept of a network

As the project progressed, the need to depart from the original working definition of a clinical network became apparent. A new definition was needed primarily to reflect the changes in the networks' composition from that of solely clinical membership to those comprising of both healthcare professionals and patient representatives.

Table 1: How the networks formed



## Network of expertise for Hypomelanosis of Ito

### HITS Support Group: Strategic Aims and Objectives

- To enrich the lives of families and children affected by Hypomelanosis of Ito by facilitating and encouraging communication and linking families together, and by being a focal point offering verbal and written support.
- To put families who have a common situation or difficulty in touch with each other to enable them to gain mutual support and friendship.
- To reduce the sense of isolation families and children with disabilities or special needs often feel.
- To liaise with the medical profession to communicate the group's existence and to share medical information.
- To promote the importance of the group and the need for communication.
- "A long term future aim that we would like to achieve is to organise a medical training awareness day in an attempt to gain earlier diagnosis." - Terri Grant, Chair.

The HITS network objectives were informed by the overall goals listed above. Two consultant dermatologists that had an existing relationship with the patient group were identified as Hypomelanosis of Ito (HI) specialists. However, when invited to act as the clinical leads for the network, situational factors imposed limits on their level of involvement.

The lack of clinical expertise and awareness of the condition within the medical community resulted in network activity being heavily reliant on the input from its members. These individuals could most appropriately be referred to as expert patients.

"Individuals become experts as they learn to cope with their chronic conditions...with proper support, people with a chronic condition can take the lead in managing their condition. This helps to improve their health and quality of life, and reduces their incapacity."

### Network objectives and activities

The primary objective of this network was to utilise expert patients' knowledge of managing HI and their experiences of navigating health, social and education systems to access appropriate support.

The project officer conducted patient interviews and facilitated focus groups with expert patients who advised on the content and overall design of the resource. Where medical information was detailed, the lead clinical dermatologist provided content and oversight.

### Impact of work undertaken

Developing the HITS patient information resource served to empower expert patients through their involvement in the information gathering process. Aimed at an audience of newly diagnosed patients, the information made available in the HITS patient resource will help alleviate some of the stress associated with the search for support services and accurate medical information that these individuals often experience.

The HITS support group has disseminated the resource amongst its existing membership base and intends to provide copies to new members as they join. Key dermatology clinics have been contacted and electronic and hard copies of the patient resource made available to them.

## Network of expertise for Ataxia UK

### Ataxia UK: strategic aims and objectives

- Finding treatments and a cure: Ataxia UK is committed to finding treatments for the different types of ataxia and to ensuring that they are delivered. To do this we recognise that we must work in partnership with researchers, clinicians and those with ataxia both in the UK and across the world.
- Treatment and care: We seek to maintain and improve standards of medical health, social care and the welfare of people affected by ataxia.
- Raising awareness: We raise awareness of ataxia among medical and social care professionals, policy makers and the general public.
- Services: Ataxia UK are instrumental to the development of services that meet the needs unique to people affected by ataxia, or that can only be effectively delivered by Ataxia UK. They continue to signpost to existing generic services which are more relevant to issues not specific to ataxia.

### Network activities

From the initial consultations held with the Ataxia UK research team, it was clear that the support group had already begun to strengthen their links with key medical professionals known to be experts in the field of ataxia. As a result, the support group had accredited two ataxia centres of excellence in England and was in the process of accrediting a third. In light of the charity's advancements in this area, the rationale underpinning the network's activity was focussed on informing the medical community of the services and expertise the support group was involved in and could refer them to.

Following extensive discussions with the patient support group and members of Ataxia UK's Medical Professional Board, it was agreed that the network would coordinate a study day for healthcare professionals known to be part of the care pathway for ataxia patients.

### Impact of work undertaken

The Ataxia Study Day provided a prime opportunity for the charity to raise the profile of its work, particularly highlighting its integral role in supporting the ataxia specialist clinics.

The content delivered by leading ataxia specialists illustrated how developments in clinical research were driving new approaches to managing the condition. Healthcare professionals in attendance gained considerable insight into ataxia and reported leaving the day with greater understanding of the variability within the condition. Many also expressed keen enthusiasm to use this knowledge within their respective clinical settings to improve overall patient care.

The success of the day demonstrated the effectiveness of using this learning method as a means of communicating the healthcare needs of ataxia patients to health professionals. This model has since been adopted by the support group, with another study day being delivered at another location.

## Network of expertise for Rasopathies

### Neuro Foundation: strategic aims and objectives

- **Inform:** We provide accurate, relevant and up to date information that explains the conditions of NF1 and NF2, the implications, challenges and opportunities, in language understood by all.
- **Advise:** We provide practical and emotional advice to help people find the best services and support available. We enable people to make choices about how they approach and live with neurofibromatosis.
- **Advocate:** We aim to increase knowledge, awareness and understanding of NF, and will campaign for change in areas we believe will improve the lives of those affected by neurofibromatosis.
- **Connect:** We aim to take a pivotal role in bringing together groups of people interested in neurofibromatosis to maximise the support and information available to them.
- **Fund:** We seek to make funds available for small projects to bodies and patient groups where we believe it will make a difference.

### Network activities

The Rasopathies network was initiated by the founder of the International Costello Syndrome Support Group (ICSSG) and two clinical geneticists based at St. Mary's Children's Hospital (Manchester).

This network was unique in its composition in that it embodied four patient support groups (INCSSG, Noonan Syndrome Association, The Noonan Syndrome Support Group & the Neuro Foundation) representing three distinct genetic conditions (Costello Syndrome, Noonan Syndrome & NF1) denoting a class of developmental syndromes caused by a common germline mutation (MAPK pathway).

The network's collaborative efforts were galvanised by its commitment to support affected families through promoting research designed to improve the understanding of all aspects of the conditions and ultimately aid the development of treatment trials.

As a means to achieving their goals, the network's primary objective was to raise the awareness of the group of conditions. It was agreed that an umbrella campaign by the group to raise awareness amongst the general public was more likely to lead to confusion, so the network did not pursue this any further. As awareness of the three conditions amongst the medical community was found to be comparatively low, patient representatives agreed that targeting this audience would be most appropriate for a networked approach, whilst remaining of benefit to their members. The network adopted a two strand approach:

1. **House of Lords reception event:** this provided a prime opportunity for patients and clinicians to meet. The occasion attracted a delegation of 120 patients, healthcare professionals and policy makers whose attendance signified their support for the launch of this clinician-patient partnered initiative.
2. **Study day for medical professionals:** as clinicians are required to attend regular training sessions as part of their medical practice, network representatives agreed that this was a suitable format for healthcare professionals to learn about the research developments that were rapidly translating into healthcare practice. 80 healthcare professionals attended the day where expert

clinicians delivered information relating to the diagnosis and natural history of these conditions, the role of molecular genetic testing and emerging treatments.

### Impact of work undertaken

The House of Lords reception event was a catalyst for the network's US counterparts who, soon after attending the event, formally recognised their attempts to mobilise support groups representing respective patient support groups in the USA. These international communications raised the profile of the network within the UK and within Europe.

Feedback from the medical study day was extremely positive with healthcare professionals expressing their interest in attending future events of a similar nature. Both clinician and patient representative network members recognised the success of the event and its cost effectiveness. As a result of the feedback obtained, network members have begun researching alternative methods of funding this work in other locations within the U.K.

### Study day checklist

**Audience:** Identifying your target audience is key. Who are you seeking to educate and/or influence? In order to make sure running the day is viable, establish the minimum number of attendees to ensure running the day is cost effective. Have a communication strategy. Utilising specialist forums will help to promote the event amongst your target audience. As a general rule of thumb you should advertise the event on websites, and within publications of specific Royal Colleges, Associations, and/or Societies to whom your desired audience are expected to hold membership with.

**Programme:** It is important that the content covered during the sessions reflect the key issues as expressed by patients whilst remaining topical and of clinical relevance to the core of the healthcare professionals in attendance. This can be achieved by having a working group consisting of patient representatives and clinicians to inform the development of the programme.

**Location:** An educational day for medical professionals should be held in a venue that is easily accessible – perhaps a hospital or university within, or near a centre of excellence for the condition/s.

## Network of expertise for Familial Adenomatous Polyposis

### FAP Gene UK and PolyPeople: strategic aims and objectives

FAP Gene UK and PolyPeople aim to provide support, information, and education to people whose lives have been affected by Familial Adenomatous Polyposis (FAP) and Gardner's Syndrome, Peutz Jegher's Syndrome, Juvenile Polyposis and MYH (Mut Y Homolog). Although the two groups approach these goals with considerable difference, their objectives serve to:

- Raise awareness of FAP.
- Support and signpost affected people to sources of information that might offer some guidance.
- Promote and maintain close relationships with members of relevant stakeholders.
- Raise funds for St. Mark's Hospital Polyposis Registry, where and when possible (PolyPeople only).

### Network activities

Led by two specialist clinicians and one representative from each patient support group, this network's activity was focused on generating and collating the evidence base needed to assess of the rationale for establishing and maintaining specialist services for FAP patients.

Working in partnership with key stakeholders, this process facilitated important consultations where various models of delivering patient centre care were discussed, and created a schedule for ongoing dialogue between interest groups.

### Impact of work undertaken

The data generated by the focus groups and meetings provide an invaluable foundation for future research. As it stands, the information obtained holds immediate translational value and has been disseminated amongst relevant healthcare professionals and commissioning bodies.

The following points in the two sub sections entitled "Factors facilitating delivery of care to patients with FAP" and "Barriers to delivering effective care to patients with FAP" were highlighted by Miss Sue Clark (general surgeon at St Mark's hospital, London).

### Factors facilitating delivery of care to patients with FAP

- Multidisciplinary expertise – paediatric gastroenterology, gastroenterology, specialised endoscopy, genetics, surgery, psychology services all co-located. This can happen because we are seeing a critical mass of patients with FAP.
- Specialist nursing team (nurse endoscopist, nurse practitioners, nurse specialist) allows the development of a high level of expertise and continuity of care for patients and families.
- Robust data collection and patient recall system (stand alone) underpins clinical management, quality control and research.
- Co-existence of research and a busy clinical FAP practise allows high quality research to be undertaken, which in turn ensures that clinical management is up to date and evidence based. The patients have access to clinical trials.

- Dedicated administrative team allows manning of telephone and e-mail helpline, production of information leaflets for patients and clinicians, maintenance of website and running of annual patient information day.

#### Barriers to delivering effective care to patients with FAP

- Fragmentation of NHS services and poor education/information resources for clinicians mean that referral to this specialist service is haphazard.

#### NHS commissioning and planning process:

- Encourages competition between units, rather than cooperation to allow a small number of sufficiently sized units to manage this (and other) rare conditions.
- Encourages local provision of care for common conditions, rather than nationwide provision for rare conditions.
- Emphasises new follow-up ratios, resulting in significant financial penalty for institutions caring for chronic long term conditions such as FAP where long-term hospital based follow up is undoubtedly required.
- Blanket policies (such as Trust discharge policy – discharging patients who do not attend one or two follow-ups) are not appropriate, and result either in long term morbidity for patients and at risk relatives, or confusion and a large amount of administrative input to rectify.
- Inadequate IT infrastructure and support to allow robust recall and data collection. We have supplied and run our own system.
- Research funding is contingent upon the disease in question being common, resulting in potential large scale benefit. This hampers research into rare conditions.

## Network of expertise for Cerebral Cavernous Malformations

### Cavernoma Alliance UK: strategic aims and objectives

Cavernoma Alliance UK (CA UK) is a charity created by people affected by cerebral cavernous malformations (CCMs). Their mission is to improve the quality of life for individuals, carers, and families affected by the condition and seek to do this by providing the following:

- Education: CA UK seeks to ensure that every person with a CCM, and their family, has access to clear information about the condition.
- Support: CA UK provides a network of support via the online forum and patient group meetings (Caverhubs) that take place throughout the UK.
- Promotion of research: CA UK has partnered with academics and clinicians undertaking research into CCMs and will continue to do so.

### Network activities

The network of expertise for CCMs was formed by CA UK and a clinical interest group consisting of three specialist clinicians nominated by the patient support group and ratified by their professional governing bodies. The network was created for the sole purpose of scoping the viability of producing clinical guidance for the care of patients affected by CCMs. Once the clinical leads had assimilated and reviewed the available research evidence on current best practice, it was agreed that this work should be extended to the creation of guidance identifying standard measures to evaluate clinical effectiveness and record patient outcomes.

The work was a more substantial undertaking than originally anticipated and thus the project team commissioned a larger (network) research group. Clinical leads engaged in consultation and peer review processes, which included patient participation from CA UK.

### Impact of work undertaken

Due to the dearth of available research evidence concerned with recommended patient pathways, clinical interventions, and resultant health outcomes for CCM patients, there was an inevitable limitation on health promotion advice permitted within the scope of the work undertaken. Even whilst considering such confounding factors, this work demonstrated immediate and long term benefits for CA UK.

By establishing the foundations of clinical guidelines for the management of CCM, the network has pioneered good practice for this patient population. Ultimately the patient group anticipate that this shall translate into clearer information being provided to doctors who are resistant to relying on volunteer support groups in the Third Sector. Having been made aware of the condition by recognised neurological, neurosurgical and genetic experts, clinicians and doctors may be more sympathetic to the support and information about the condition which CA UK can provide.

## Conclusion

One of the achievements of the Facilitating Networks of Expertise project has been to highlight the value of implementing initiatives which engender collaborative work between patients and clinicians. Their efforts, which have led to the production of patient resources, clinical guidelines and two successful study days for healthcare professionals must be applauded.

Whilst acknowledging the significant achievements made, the processes undergone to deliver these outcomes also offer vast scope for shared learning. They demonstrate practical and transferable models of good working practice, whilst illuminating ways in which patient organisations can contribute to the development of networks for genetic conditions.

Patient and public involvement (PPI) in commissioning services has been identified as one of the least well developed components of clinical governance, and clinical networks can often have a significant role in contributing to this process. Although the networks of expertise developed as part of this project were not exclusively clinical in composition or outcome, the procedures undertaken provide insight into effective mechanisms for patient involvement in the commissioning of services.

The use of qualitative methods at this scale generated a rich data source of patients' experiences that can be used to inform future work. Across the (combined) sample of 450 members belonging to the support groups participating in the project, the core themes underlined the need for improved access for children, adults and families with rare disorders to health and care professionals with experience and understanding of their condition. As patients identified a lack of communication between healthcare professionals, and miscommunication between healthcare professionals and themselves as contributing to delayed diagnosis and access to treatments, the experience of fragmented care, and the source of great frustration, future work in this area must dedicate its efforts to developing methods that would improve these communication processes.

Although the key drivers behind the work achieved encompassed the good will, enthusiasm and selfless determination of patient representatives and clinicians, the need to finance such invaluable work cannot be overlooked. Qualitative assessment of the anticipated added value for patients and care professionals must also consider the cost benefits of such work. This holistic approach will enable a thorough evaluation of such work, and ensure methods employed are the most efficacious and cost effective.