



THE LONDON SCHOOL
OF ECONOMICS AND
POLITICAL SCIENCE ■

Alström Syndrome:

**A Cost-Benefit Analysis of Multidisciplinary Treatment &
the Impacts of Proposed eHealth Solutions**

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Executive Summary

Alstrom syndrome (AS) is an ultra-rare genetic disorder affecting less than one in a million people, yet its symptoms are both manifold and severe. Patients may suffer from cardiomyopathy (heart failure), obesity, type-II diabetes, kidney and liver failure, and often lose their eyesight in childhood and may even go deaf. Due to the vast number of symptoms involved and the challenges posed by dual-sensory loss, innovative and collaborative treatment methods are required to best deal with the condition.

In this context, this report seeks to investigate the costs and benefits of multi-disciplinary clinics (MDC) in rare disease management in connection with AS. To this end, extensive interviews were performed with a number of patients and health practitioners at Queen Elizabeth Hospital (QEH) and Birmingham Children's Hospital (BCH), and a survey was conducted among all members of Alstrom Syndrome UK (ASUK), a patient-led charity. In a further step, we look at how these MDCs could be further developed into virtual clinics and virtual data hubs with the help of new technological developments in electronic healthcare (eHealth). Finally, taking a more holistic view, we turn to AS disease management as a paradigm for other rare diseases and how the ASUK approach to treatment could be replicated to service other charities.

Our methodology and results are as follows:

I. Cost-benefit analysis of MD clinics

Methodology: The costs and benefits of the MD clinics were considered from three angles - an institutional (NHS), a patient, and medical practitioner's perspective. This was achieved with the help of NHS costing data for the services provided at the clinics, telephone interviews with patients and a comprehensive patient survey, and face-to-face interviews with the lead physicians of the MD clinics.

Results: *MD clinics are highly valued by both patients and physicians and can be run at an affordable cost.*

In the costing analysis, the total direct variable cost of one of the quarterly MD clinics serving 6 patients was found to be GBP 4,138. It is important to note, however, that this figure only includes the direct specialists and consumables involved – i.e. the actual total cost may be considerably higher. The number was calculated as the sum of all personnel costs of the experts involved and an estimate for the consumables used in the treatments. This figure specifically excludes any non-direct or allocated costs, such as overhead costs (e.g. allocated rent), depreciation of capital goods (such as machine hours used) or organisational run-up costs.

At the same time, the benefits of MD clinics were found to be far-reaching and great. Patients commended the easy access to treatment and the niche medical consultations they were able to benefit from at the clinics. Time savings were found to be extensive and patient quality of life – both from a psychological viewpoint (more support available, less anxiety) and a socio-economic perspective (fewer work days lost, less travel between facilities) – was shown to be considerably improved.

From a practitioner's viewpoint, the "improved decision-making efficiency" at the clinics was key and the reduction in side effects thanks to coordinated treatment of multiple conditions. While the physicians conceded that coordination efforts regarding the scheduling of the necessary number of specialists and patients was great, this was found to be outweighed by the overall reduction in bureaucracy thanks to ad-hoc decision-making (due to the extensive number of specialists onsite).

II. Evolving MD clinics into virtual clinics with the help of eHealth

Methodology: Building on the needs of AS patients derived from the patient interviews and survey, the possible role of eHealth as part of a virtual clinic was researched. This research is founded on the one hand on a literature review (describing the advantages of eHealth and its current shortcomings (e.g. in relation to dual-sensory loss), and on the other hand on an extensive review of service provider offerings from IBM, Gradiant, Cisco, Vodafone and Orange.

Results: *eHealth applications have the potential to considerably enhance patient quality of life, and our competitor analysis revealed that Orange currently has the best portfolio of eHealth solutions for AS; however, pricing remains a core concern.*

Although eHealth has not yet been extensively researched and there is little empirical evidence, the literature nonetheless points out some of its key benefits. These range from patient empowerment thanks to self-management of the disease to benefits from early detection of new symptoms and conditions. However, there are still considerably hurdles to be surpassed in eHealth regarding process and data standardisation and data protection and privacy concerns.

In the analysis of the five eHealth providers, Vodafone and Orange were found to be the leading companies. Both offer a complete eHealth service from consultation and prototyping to implementation and monitoring. IBM and Cisco, were ruled out on the basis of geographical location (in the USA), and Gradiant did not have any in-house product or platform. Of the five, Orange remains the leader due to its tailor-made applications for ASUK, its complete project pricing and duration, and its competitive devices suited for the needs of AS patients.

III. Alstrom Syndrome as a paradigm for rare disease management

Methodology: In order to determine how ASUK's work could be replicated as an "off-the-shelf" business model for rare disease management, the needs of other RD charities were determined in a series of qualitative interviews with Ataxia-Telangiectasia society (A-T society), The Barth Syndrome Trust, Vasculitis UK, Meningioma UK, and Lymphangiomatosis & Gorham's Disease Alliance - Europe.

Results: *ASUK's work is highly relevant to the needs of other rare disease charities, yet these may need additional support in implementing it due to lack of resources*

The rare disease charities interviewed showed similar needs in treating their multi-symptom conditions and the vast majority of those interviewed supported the MDCs as an ideal means for treating such a condition. However, two charities pointed out that they did not have the resources to sustain the set up of an MDC and would require extra time and funding. In general, eHealth was seen as a promising trend although the need for customisation of apps to patients' needs was underlined.

Overall, there is therefore a strong case for expanding on ASUK's multi-disciplinary approach to treating AS. While the clinics themselves have proved an invaluable first step to more integrative treatment methods, further collaboration is needed – both between different medical facilities and across national borders to develop successful treatment practices.

eHealth is surely a promising development in this field; yet the challenges posed by dual sensory loss combined with the high prices currently charged by operators still pose significant barriers to its implementation. This said, these two restraining factors are bound to be resolved in the medium term as technology progresses and mobile applications become more integrated in the medical system. If/when this occurs, Alstrom will perhaps transition from a paradigm for multidisciplinary clinic-treatment to a first mover in the eHealth domain. In this case, rare disease charities all over the EU could learn from the "Alstrom Approach" in managing their own conditions.

Foreword

It has been our pleasure to compile this report for ASUK and to investigate Alstrom Syndrome from an economic angle. As LSE students, we are not medical practitioners, nor do we have any training in that area, so for any medical inaccuracies in this document we must apologise. Regarding the economics, there are a number of variables that are hard to quantify – particularly benefits to patients' quality of life, reduced anxiety and depression, or even a longer life expectancy. In our effort to analyse these factors and partly quantify them, we have had to make a number of tough assumptions that can no doubt be argued. However, in compiling this report, we have had – first and foremost – the patients' best interest at heart, and have made any such assumptions with due prudence and humility.

At this stage we would also like to thank Kay Parkinson from ASUK for her role as mentor, ever-flowing source of information and guidance in compiling this document. Moreover, it was a pleasure to work with Professor Tim Barrett, Dr. Tarek Hiwot and the whole team from ASUK and its patients. Finally, our sincere thanks goes out to Dr. Pietro Maffei and Dr. Vera Bettini from EUROWABB Italy for collaborating with us on the provision of medical data.

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Cost analysis of multi-disciplinary clinics

Multidisciplinary clinics

Birth of multi-disciplinary clinics

Individual treatment, the norm in fighting Alstrom Syndrome, proved an insupportable approach. Not only was dealing with a multi-symptom condition in various ^medical practices time consuming; patients were commonly confronted with medical staff that were illiterate in AS and therefore unable to make either a timely diagnosis or prescribe adequate treatments. Even more alarmingly, overlaps in medication targeted at different conditions in some cases led to negative (and avoidable) side effects.

For this reason, ASUK, as patient led charity promoting AS treatment and awareness, fought to set up multi-disciplinary clinics in cooperation with Birmingham's Children's Hospital (BCH) and Queen Elizabeth Hospital (QEH) for AS patients across the UK. The clinics are held quarterly and last for two days; during which patients can undergo all necessary treatments for the conditions they may exhibit.

The clinics in action

The two hospitals provide three important services for adults and children respectively:

1. First and foremost, QEH and BCH provide monitoring and treatment for all conditions of AS, with individual patients following customised schedules. Importantly for the patients, they receive immediate feedback from the practitioners and do not have to wait for results or the proper medication.
2. Second, the clinics provide an important role as information providers and hold talks and information sessions on AS for the benefit of the patients.
3. And third, QEH and BCH are growing into important knowledge hubs for AS and could therefore provide an important function in developing cures and new treatment methods. In this context, it is very important to note that for any treatment to be brought to market, it must first undergo a series of tests for which a large enough sub group of patients is needed. In rare – or ultra-rare – diseases this sample size is often hard to achieve, making patient and knowledge clusters invaluable. Moreover, there is even talk that AS research could benefit patients suffering from single AS symptoms, such as Diabetes, which might suggest that the knowledge hub function could benefit a far larger target group than merely AS patients.

As four clinics are held each year (in BCH with six patients per clinic), patients are free to choose between sessions. Common practice is to attend at least one clinic per year, yet depending on the medical condition of the patient, more regular attendance is possible

Flow chart of AS diagnosis and treatment

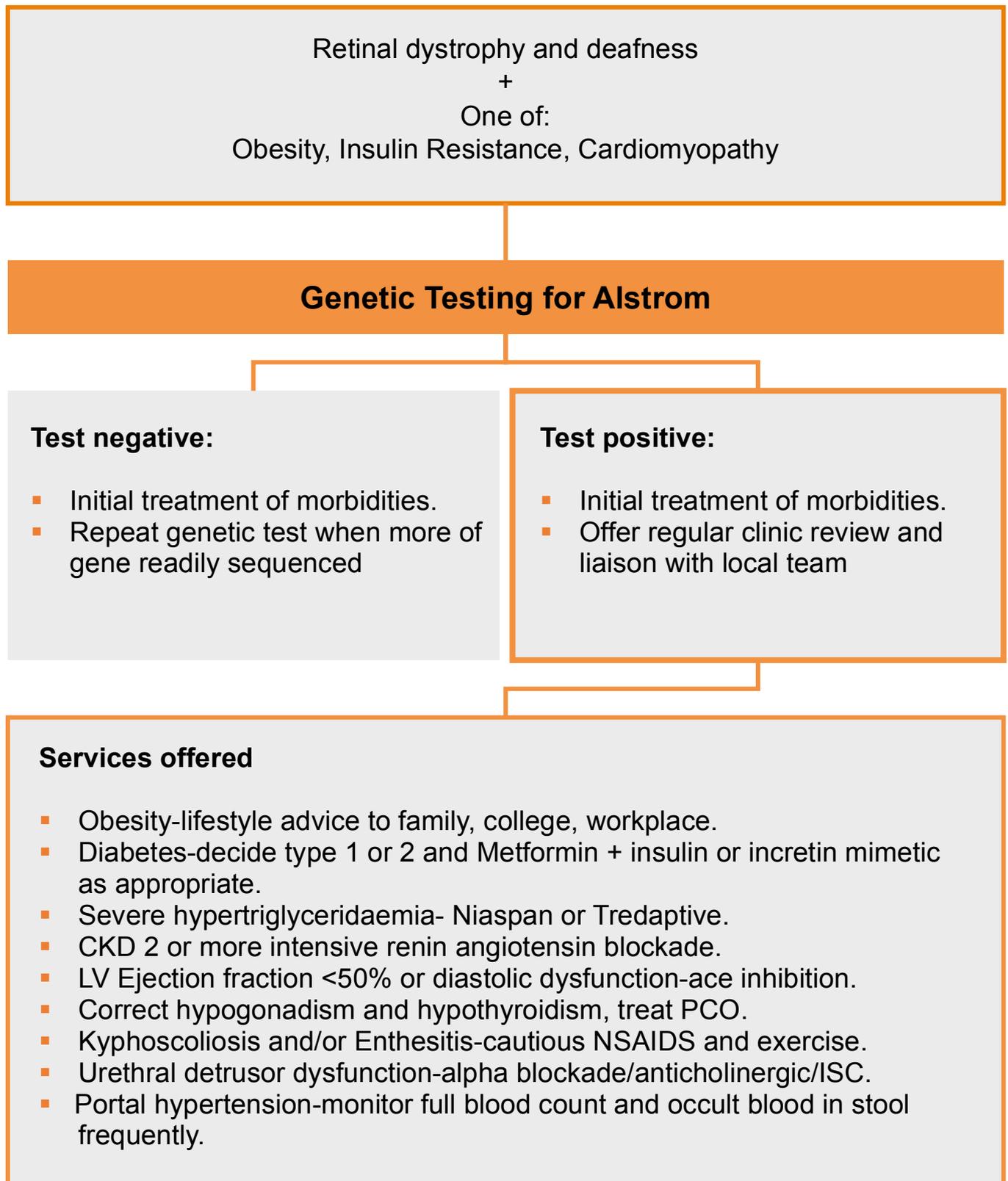


Figure 1 Flow chart of AS diagnosis and treatment (Adapted from NHS Service specification based on adult clinic in Torbay, 2010)

Structure of the clinics (based on BCH and QEH)

The clinics take place over two days in the course of which all tests are performed. There is nearby accommodation in a hotel 100m down the road (at BCH) providing convenient access, and all treatment facilities are clustered on two floors of the hospital to make the transition from one specialist to the next as easy as possible.

Initial appointment with ASUK support worker or lead physician. This appointment is scheduled in the two-month period leading up to the clinic. The objectives of the appointment are to (i) ascertain which treatments the patient needs, (ii) make sure that the patient sees all specialists required at the MDC, and (iii) to see if the patient has developed any new symptoms that may need to be treated at the MDC.

Day 1

Round table discussion with patients and lead physicians (2hrs): All patients meet jointly with the doctors and freely voice their concerns and experiences – both with the condition and AS treatment. In the adult clinic, the round table discussion occurs at the end of day 2, meaning patients can also give feedback on how the clinic went, what worked well and what did not.

Dietician (4hrs): gives advice on food, especially in relation to obesity and diabetes.

Exercise physiologist (8hrs): works on patients' movements and posture, which is often bad as patients avert their eyes and lower their head due to their sensitivity to light.

Cardiac technician (4hrs): conducts blood pressure test.

Phlebotomist: takes blood samples, which can be difficult due to obesity and child resistance.

In the afternoon, time is devoted to exercising and life style advice.

Day 2

Cardiology (4hrs): Discussion of test results and monitoring of the heart

Diabetes specialist (4hrs): Consultation on life with diabetes and blood sugar levels

Respiratory medicine (4hrs): General consultation

Endocrinology (8hrs): tests relating to internal organ functioning

Ophthalmology (8hrs): consultation and treatments for eyesight loss

Psychologist (4hrs): Consults on common feelings of anxiety and depression

Genetics counsellor (4hrs): Undertakes genetic analysis and risk investigations in family relations

Finally, it should be noted that there are a number of minor differences between the children and adults' clinics that are elaborated on in the next section. These differences notably concern the timing of individual sessions and the duration of the event.

Costing of MD clinics

Fundamental premises

To better understand the efficacy of the clinics, it is necessary to understand their broad cost structure – both on a per-clinic and a per-patient basis. This achieved, direct comparisons between MD clinical treatment and other treatment practices can be drawn. For example, a government wishing to know the monitoring and treatment costs of all AS patients in a country could use this data to estimate the efficacy of single treatment vs. multi-disciplinary treatment from a cost perspective. What is more, this pillar can be expanded on to include new treatment methodologies, such as eHealth and virtual clinics. However, it should be noted right at the start that the figure reached is an approximation and no more.

Summary of approach

In a first step, we set up a detailed overview of all aspects of the clinics with the help of patient interviews. In a second step, we conducted physician interviews to gain a more detailed (and medical) picture of the exact treatments involved, their layout in the hospital and the respective personnel involved in each practice. Third, we then spoke to a finance manager from the NHS who was able to provide valuable insights into the costs involved in each treatment (better: the cost categories employed by the NHS, which were important as they determined the format of the data we could input into our model). What is more, we were able to obtain hourly rates for the experts involved (using middle-of-the-bandwidth pay scales in each case) and multiplied these rates by the hours each person was likely to be involved.

Based on our initial interview findings, we therefore had three cost categories:

1. **Variable direct personnel costs** defined as the hourly rates of specialist physicians (such as a cardiologist), technicians (such as a cardiac technician), nurses (for example for monitoring purposes) and support personnel (for example providing organisational assistance and helping moving patients between rooms)
2. **Consumables** are defined as all materials that might be used in a treatment that are non-capital expenses, and would mostly be used only once (such as bandages). In the model, these were taken as a percentage of the personnel costs and adapted to the best of our knowledge depending on the treatment type (for example, a psychologist arguably does not use any consumables, whereas an ultra sound examination would).
3. **Capital expenses and overhead** are found to be two-fold: first, there is the capital equipment to consider that is used to perform the examinations. If this equipment has a useful life of 5-10years, for example, this cost could arguably be allocated to each treatment. However, as there is no direct association between the use of the equipment and their depreciation, this cost was not assigned to the MD clinics. We believe this is particularly fair as to the best of our knowledge, no additional equipment had to be purchased to run the MD clinics and all equipment was already available and in use. The same logic applies to overhead costs, such as rent. As no additional facilities had to be rented to run the clinics, this cost is arguably fixed and does not need to be considered in the model, which in theory should only investigate the marginal cost of treatment.

Limitations to data and data quality

While some of the variables involved are likely to be quite accurate (such as the specialists' pay data), others are based on some educated assumptions and others still – such as indirect costs and run-up / follow-on costs – are not included, making the estimate susceptible to undervaluation.

First, this relates to the hours each physician *really is* involved in the treatment, which may well deviate from the nominal hours on paper that person is said to be involved. In practice, this may mean that the diabetes specialist is booked for 4 hours but, in fact, requires 6 hours to see all patients. Also, any costs incurred running up to or following up on the clinics, e.g. for organisation r follow-ups, are specifically excluded. This means that the actual cost is likely considerably higher.

Second, the cost of consumables could be criticised to be too vague for astute scrutiny. This is undoubtedly true, as we have no observations to back up our assumptions. However, it should be noted that the personnel costs are by far the major component of the MD clinical cost, making the impact of the consumables less strong.

Third, it should be noted that we, as LSE students, did not have access to many tools, contacts, and databases that would normally be available to professional NHS costing staff. We ran into considerably hurdles in our search for data, and while we are pleased with the result, we fully acknowledge that with the right resources and considerably more time, the picture of the MD costs could be improved. This relates in particular to the treatment costs of Alstrom patients outside the MD clinics, which we were not able to include in our model for lack of resources, but would in fact be a vital component to the accuracy of our calculations.

Results

Based on the physician interviews we were able to identify 12 areas of treatment, which we bundled into 12 cost centres. Each cost centre was assigned both its direct variable personnel costs and the cost of consumables (if any). As stated above, capital expenses and overhead were not allocated.



Based on this analysis we can identify major and minor cost drivers

Major cost drivers

(>10% of total cost)

- Cardiology (12%)
- Ophthalmology (18%)
- Endocrinology (14%)
- Assisting personnel (26%)

Minor cost drivers

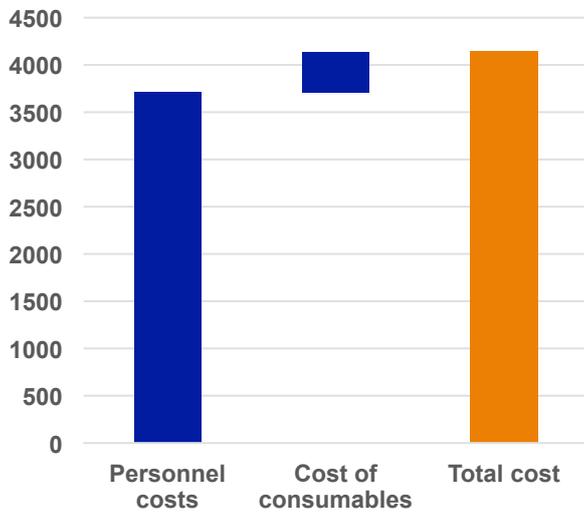
(<10% of total cost)

- Pre-MDC counselling (3%)
- Genetics (9%)
- Morning group discussion (3%)
- Diabetes (2%)
- Dietary advice (2%)
- Physiotherapy (5%)
- Psychology (2%)
- Geriatrics (4%)

Figure 2 Major and minor cost drivers

The total cost figure of GBP 4,138 per clinic can be further broken down into its personnel cost component and the cost of consumables involved

MDC cost broken into personnel and consumable costs



The cost analysis of the MDC hereby gives us an indicative estimate of the costs involved in the set-up, running and treatments involved in the MDCs. As pointed out in the data limitations section, though, much deeper analysis of the respective services involved would have to be conducted to make this figure more realistic.

Figure 2 MDC by cost type

MDC cost drivers

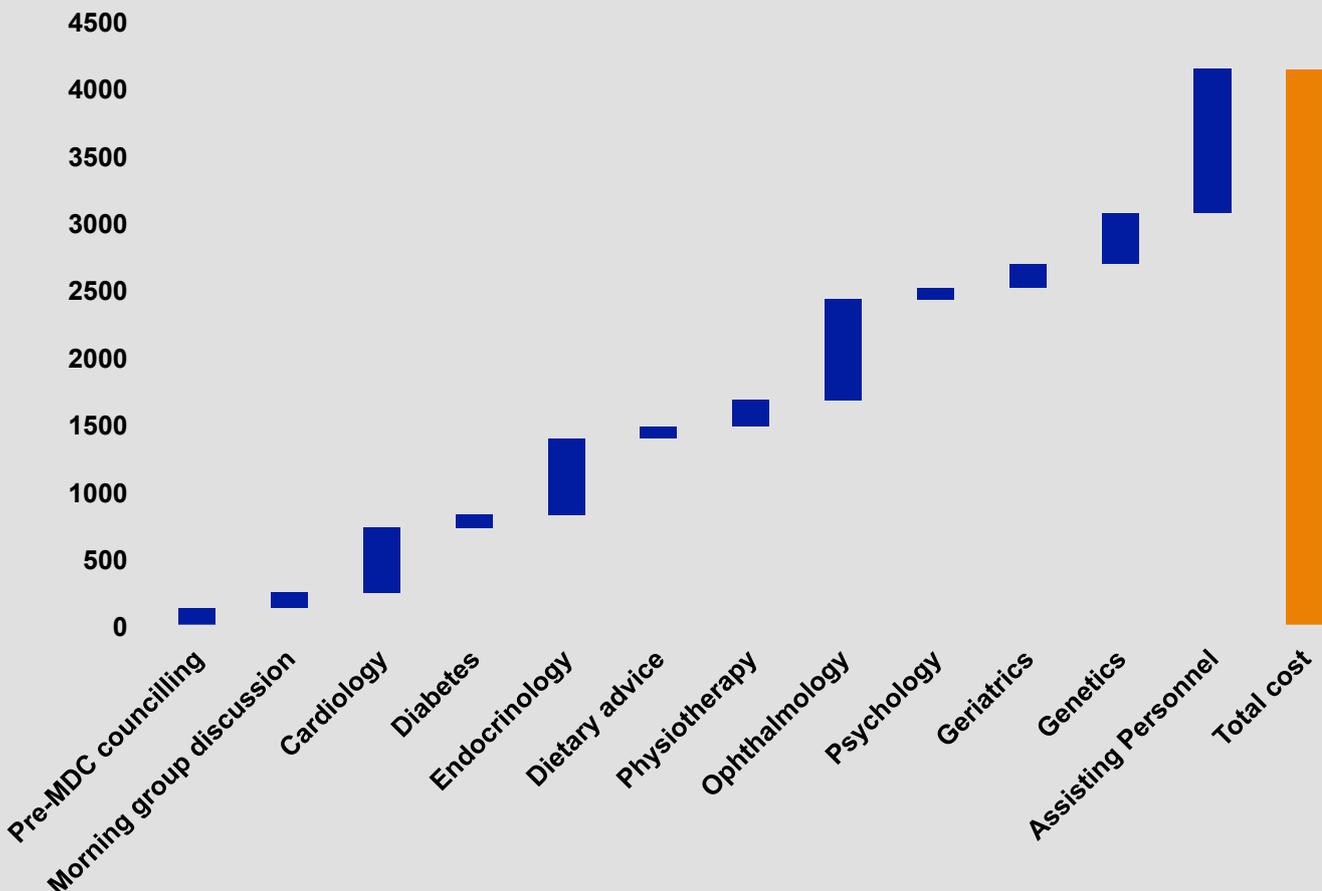


Figure 4 Cost drivers of MD clinics

Survey results on multi-disciplinary clinics

Analysis of Alstrom Syndrome Survey

Insights from patients and caregivers

Up until this stage our analysis has been dominated by quantitative evaluations of the direct costs associated with the resources used in the medical treatment and management of Alstrom Syndrome. This was used to quantitatively describe the benefits derived from the introduction of multidisciplinary clinics. However, an important part of our analysis is to also demonstrate the indirect cost benefits associated with the clinics.

We conducted a multi themed questionnaire survey to shed some light on these indirect costs. The survey provided us with a rich set of statistics that demonstrate some of the indirect benefits of multidisciplinary clinics (outside of medical treatment costs that have traditionally been the focus of most cost benefit analyses). The data we received ranged from quantitative statistics to patient led long answer feedback. Overall, our findings can be extrapolated to the wider rare disease community and be used to improve our understanding of the impacts rare diseases have.

For more information regarding our survey approach, design and methods of data capturing please see:

Exhibit 1: Approach and Exhibit 2: Methods and Data Sources.

There are roughly 60 families in the UK that are suffering from Alstrom Syndrome. Alstrom Syndrome is one of approximately 7000 rare diseases that have been identified across the globe. When compared to widespread conditions, rare diseases attract far lower degrees of attention. Yet collectively rare diseases have an expansive impact. In the UK alone there are 3.5 million people living with rare diseases. Around the globe the rare disease community is estimated to include 350 million people. Given the fact that rare diseases impact more than just the patients, it is clear that a greater degree of understanding of them could have far reaching implications. As with most rare diseases, Alstrom Syndrome care journeys are predominately characterised by misdiagnosis, conflicting medical opinions and stress. As such any success solutions introduced by Alstrom Syndrome institutions have the potential to act a paradigm for other diseases and the ability to act as a catalyst of change and improvement.

The overarching themes we attempted to address with our survey included:

- Career implications for patients and caregivers as a result of Alstrom Syndrome
- The resulting benefits of multidisciplinary clinics over individual treatments not highlighted by quantitative cost models
- Morbidity costs associated with the treatment of Alstrom Syndrome, particularly travel implications and work hours lost
- The common theme of misdiagnosis, conflicting medical opinions and stress
- The lack of information sharing and cooperation required to address and diagnose Alstrom Syndrome
- The economic, financial and emotional toll Alstrom Syndrome has on patients, care givers and their families
- Insights into patient and caregiver opinion, attitudes and views toward proposed eHealth solutions.
- Determining the lifetime earnings lost to the economy due to the premature mortality of Alstrom Syndrome patients.

The survey resulted in the following sets of results:

- Quantitative statistics demonstrating what life is like, living and managing Alstrom Syndrome as well as some of the significant benefits generated as a results of ASUK's multidisciplinary clinics.
- Qualitative data providing meaningful insights into some of the indirect costs associated with Alstrom Syndrome and patient feedback on multidisciplinary clinics
- Opinion, attitudes and views toward proposed eHealth solutions.

Analysis of Alstrom Syndrome Survey

Findings Call for the Following:

- **Increased attention paid to Alstrom Syndrome, from a legislative, policy and economic perspective** – *The results of our survey point toward the profound economic and financial impact Alstrom Syndrome has on not only the individual and their families but on society as a whole.*
- **Increased collaboration amongst physicians and improved awareness of the diagnosis criteria of Alstrom Syndrome** - *Improved awareness, educational programs, networking opportunities and better collaboration point toward better treatment practises and more efficient and effective diagnoses*
- **Increased information sharing as the pivotal driver of research**
- **Continued flow of resources dedicated toward dealing with the emotional impact of living with Alstrom Syndrome** – *There is a tremendous emotional burden associated with Alstrom Syndrome. Any resources that assist in navigating the process are warranted.*
- **The promotion of multidisciplinary clinical treatment** – *Results from our survey point toward clear financial, economic and emotional benefits as well as improvement in diagnosis, information sharing, educational awareness and treatment of the syndrome.*
- **A clear case of community support for the introduction of eHealth solutions** – *The survey revealed a clear avocation of eHealth solutions and belief that they would make management of Alstrom Syndrome more effective and improve the overall quality of life.*

Theme: Patient & caregiver sample overview and career implications

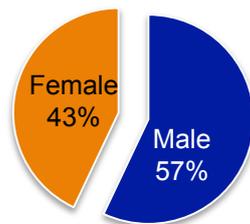
The survey was distributed by way of the charity organization Alstrom Syndrome United Kingdom (ASUK) via their database of Alstrom Syndrome patients.

According to our survey we identified the following results:

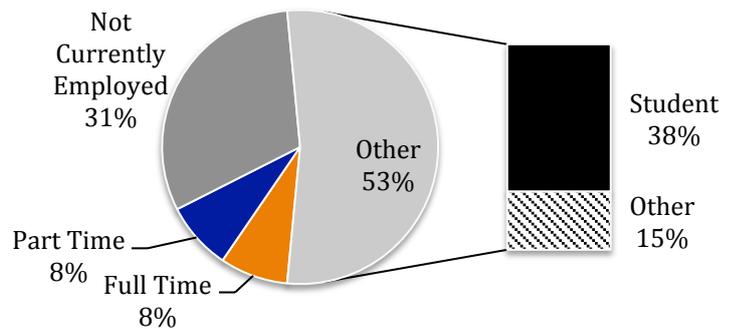
Patient overview and career implications:

22
Average age of Alstrom Syndrome patients surveyed

Patient respondents' gender



Patient by Employment Type



14%
of patients had to leave their previous occupation as a result of the Alstrom Syndrome condition

73%
of patients who said they had sacrificed opportunities for employment as a result of Alstrom Syndrome

Examples of previous occupations:

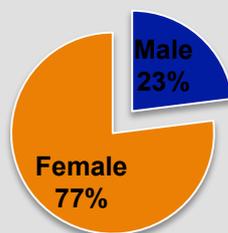
- Area Banking Director
- Volunteer
- Trustee of Ciliopathy Alliance Ltd
- Administrative Assistant (Audio-typing)

Caregiver overview and career implications:

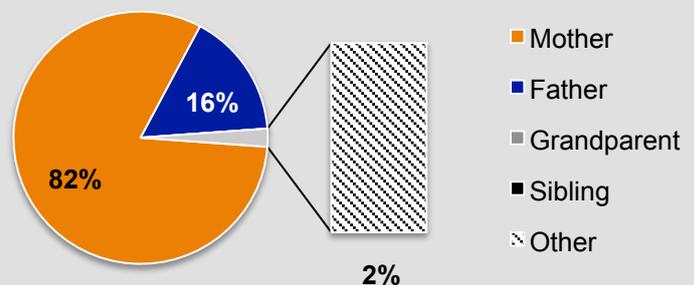
Often, when dealing with illnesses such as Alstrom Syndrome results in patients and caregivers alike, leading very different lives than what they expected. Caregiver are often required to become full time carers and find themselves juggling multiple roles in an effort to receive optimal care.

45
Average age of Alstrom Syndrome caregivers surveyed

Caregiver respondents' gender



Caregivers relationship with patient



57%
of caregivers were required to become full time caregivers

46%
of caregivers that were required to reduce the number of hours spent at work or had to seek more flexible employment to work around the needs of the person with Alstrom Syndrome

Examples of caregiver careers:

- Lawyers, Accountants, Nanny, Gas Engineer, Geologist, Managers, Mechanics, Shop Assistants, Family Support Officers, Nursery School Manager

Key insights into the patient and caregiver career implications

- Patients still try to enjoy their life and have ambitions. However, often “career perspectives” have to be given up due to the syndrome.
For example, one patient was a high street banker close to directorship before being diagnosed with Alstrom Syndrome. The severity of his symptoms meant he had to forgo his career.
- As for caregivers, taking care of patients can pose significant burden. The father of one patient said that he is still working in a 50-hour-per-week job, which is really stressful and almost impossible to handle because he has to take care of his son.
- According to patients and caregivers, the introduction of MDCs has reduced the time required a lot by concentrating treatments on single days.

Theme: The role of ASUK’s multidisciplinary clinics in treating Alstrom Syndrome

ASUK’s multidisciplinary clinics have had a number of profound benefits in relation to the management of Alstrom Syndrome. Our survey revealed a clear patient led backing for the clinics and the support structure implement by ASUK. Clinics have played a key role in reducing morbidity costs associated with the syndrome, in relation to both more effective treatments as well as reduced intrusion of the syndrome on everyday life. The number of visits to physicians are drastically reduced which in turn minimises the number of days away from everyday life that have to be committed to management of the syndrome. Our survey revealed that patients require fewer visits to individual hospital appointments, quality of life has improved, there is a higher degree of support offered, services are more patient led and better coordination of research efforts are enhanced.

According to our survey we identified the following results:

9.5

Average rating patients gave to the importance of the support offered by the multidisciplinary clinics.

18%

of patients with AS that felt the health care institutions before the establishment of clinics, provided sufficient resources to properly treat the syndrome

1.3x

Average number of times patients attend ASUK multidisciplinary clinics annually

17 days

Average number of days of treatments/evaluations/test required completing annually to manage AS **before** clinics

7 days

Average number of days of treatments/evaluations/test required completing annually to manage AS **after** clinics

Qualitative Feedback on the Role of ASUK's MD-Clinics

– Patients felt they had to attend fewer individual hospital appointments

“

Yes I attend 3 clinics a year and I feel that all my needs are fulfilled from these clinics. I only have one other hospital to go to which is an eye hospital, which I feel is an acceptable amount of extra appointments

Without question I have far less outpatients appointments to attend which is obviously to me a time and cost saving

Yes - we have all the tests over two days and the results are given before we leave. We then know when we need to return.

Since the introduction of the AS clinics it has really helped manage the appointments. It gives me something to latch onto which I can seek advice and guidance from QEH about any health related matters.

Yes feel more happy and confident that my child is well looked after

ASUK have through their booklets helped the other hospitals I visit to treat me with better care and give me the right drugs, blood and other tests etc.

”

– Patients felt that the quality of life had improved since the introduction of the clinics

“

Yes definitely - meeting other parents, and professionals is fantastic

The condition is life changing, and the symptoms and disabilities attached to it, are life changing also. The clinics and the professionals working within it give support and guidance

I feel better in myself and I feel confident that I only have to phone my doctor at QEH and I know I will be supported by doctors who know the condition well

Yes- although the clinics are intense we know we are seen by specialists and that they know Alstrom Syndrome and can offer expert advice

Yes. I am able to meet others who have the same condition as myself as well as learning the best types of exercise and eating plan I should be on

It is reassuring to have consultants who are more expert in understanding AS than others who see fewer patients

It has helped to get more information on Alstrom and useful in sharing our experiences. Since my child has been diagnosed it has really helped in seeking support and assistance from lots of institutions

I feel that we have a good support network

”

– Patients felt that there was a greater degree of support as a result of the clinics

“

I'm supported by personal help either online or by telephone and whenever, nothing is too trivial - before I had no support

The fact that they are all there under one roof at the same time is amazing. This makes sure that all the tests get done. The staff also has an understanding of our condition

Yes very much so, from all the medical staff whether in a supporting role or as a consultant nothing is too much trouble. Time is never an issue and you have a chance to ask questions but your family

Yes I feel like I have much more support for my daughter and us as parents. We feel that we have more people to fight [in] our corner, which helps with worry and anxiety.

I know I only have to phone a member of the team and there will be someone who will listen and help me with any problems.

ASUK are fantastic. They are mums or dads themselves and know what we are going through- I felt hope the first time I met them, members are also very much included.

”

Qualitative Feedback on the Role of ASUK's MD-Clinics

– Patients felt that multidisciplinary clinics result in a better patient led service

“

Now [we] have full support, all my needs are catered for, advice is freely given and explained in a terminology I can understand and people are friendly and not patronizing

I feel it is better after the clinics were introduced as I felt that I was being seen by specialists who knew the condition and knew how to look after me. Patients had the input of what they wanted to know and the type of tests that they felt were important and concerned them and these concerns were actually listened to.

The whole ethos of Alstrom Syndrome UK is that it is a patient led organization. Everything that is done is for the patient and close family whether that is research into treatments for a better quality of life or just someone to talk to and share experiences.

Patients know what they need- before the clinics people told us what they thought we needed

Organization is fab. Fewer days off work. Meet other people with the condition.

”

– Patients felt that charities like ASUK aid in the development of research

“

They are instrumental in encouraging and supporting research into the condition even managing and coordinating research

ASUK are an example to follow [for] other charities. They have achieved lots for families living with AS. Just meeting other parents and patients living with AS is phenomenal - we have so much in common, yet so different.

They go to events around Europe saying what AS is, that ALMS1 the defective gene causes it and they invest in research for a possible cure or partial cure and provide surveys to aid research and give confidential information to only trusted parties

They provide relevant information and support and materials such as a folder with contact details, information for school etc.

Without them we would have nothing- they have been the drivers for better care and treatment.

ASUK and in particular it's founder Kay strive daily to not only raise awareness but to search for potential treatments for what is a multi organ condition

”

– Summation of other benefits patients felt multidisciplinary clinics provided

“

We get to meet other families with the same condition and can support each other

I am very much actively involved with the charity as a member of the board, so I feel part of a unique family and I am very proud to work with people like Kay, Kerry, Catherine, John and Iram. I am also very grateful to Tarek for finally after many years getting me a diagnosis and to every member of his team for there care and support to each of us.

We appreciate the clinics will lead to greater understanding of the condition which will help all patients

Meeting other mums- high-class treatments-peace of mind that the doctors know what they are talking about

Support in social care, education, referrals to other institutions etc.

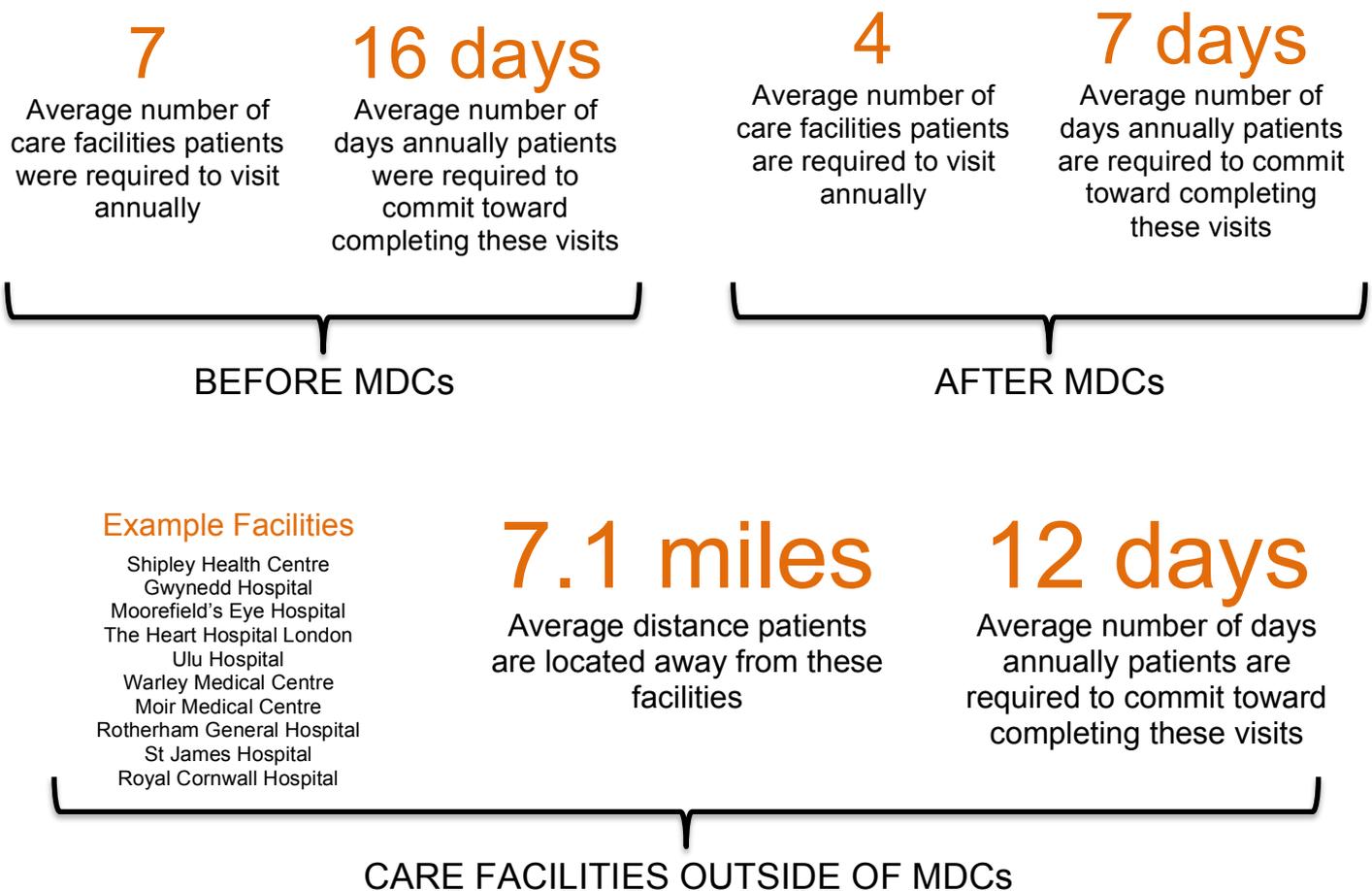
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Theme: Morbidity costs - Travel implication and work hours lost

As would be expected sufferers of Alstrom Syndrome, due to their respective symptoms, experience a loss or impairment of their ability to work as well as to engage with leisure activities. We used our survey to generate average results of our population in conjunction with nationally available statistics to demonstrate both quantitatively as well as qualitatively the morbidity costs associated with living with Alstrom Syndrome.

According to our survey, we identified the following results:

Before multidisciplinary clinics, traveling was a significant burden on patients and their caregivers



Summary of Care Facilities Before Multidisciplinary Clinics

- **35 miles** - Average distance for patients ordinary place of residence
- **5 times** - Average number of times patients were required to visit each individual facility
- **13%** - of patients were required to stay in paid accommodation during these visits
- **1.25** - Average number of nights accommodation required at time
- On average accommodation was paid for by:

20% Patient 7% ASUK 0%NHS 0% Other 73% Not paid accommodation

Key insights into the role ASUK's multidisciplinary clinics have played in the treatment of Alstrom Syndrome, including implications in terms of travel and work hours lost

Overall in the interviews with patients and caregivers as well as physicians, it stood out that patients really praise the MDC. The MDCs make their life easier:

- It is two days out of the patients' lives as opposed to countless visits to physicians.
→ TIME SAVING
- Patients benefit considerably from a fixed treatment schedule.
→ PLANING SECURITY
- Patients praised the clinics for providing "black and white" answers. Patients and caregivers just want the truth and want to know exactly what they are facing.
→ CERTAINTY
- The MD also play an important role in teaching the patients and their caregivers about the disease and the symptoms. → EDUCATION
- The comfort for patients in the MDC is higher. Due to their limited mobility they appreciate that treatments take place in one hospital and that the different rooms are very close. Also the hotel is very close to the hospital. → COMFORT

Also from a physicians' perspective the MDC offers a lot of advantages:

- Improved efficiency in decision-making due to direct communication between physicians instead of sending emails. → DECISION EFFICIENCY
- Reduced side effects due to better communication between physicians
→ FEWER SIDEEFFECTS
- From an physician's perspective the MDC has the advantage that less bureaucracy is required in treating the patients due to shorter ways of communication (e.g. can just talk to other specialist); however in planning the MDC more coordination between hospitals and physicians is required. → LESS BUREAUCRACY, but MORE COORDINATION required
- The expertise available to patients is maximized thanks to the eight specialists onsite.
→ EXPERTISE



Prof. Tim Barrett
Birmingham Children's
Hospital



Dr Tarek Hiwot
University Hospitals
Birmingham

Theme: Diagnosis history and the common theme of diagnosis delays

Most Alstrom Syndrome care journeys are characterised by delays in diagnosis. Not only does this result in an incredulous amount of resources wasted, but it also causes a tremendous amount of emotional turmoil for both patients as well as caregivers.

According to our survey, we identified the following results:

15

Average age at which patients with Alstrom Syndrome were diagnosed

2.5

Average number of misdiagnosis before being diagnosed with Alstrom Syndrome

More than 20

Average number of visits to physicians before being diagnosed with Alstrom Syndrome

57%

of patients that reported their diagnosis was as a result of their own personal investigations into the possible cause of symptoms

Summation of survey respondents' symptoms (% of sample)

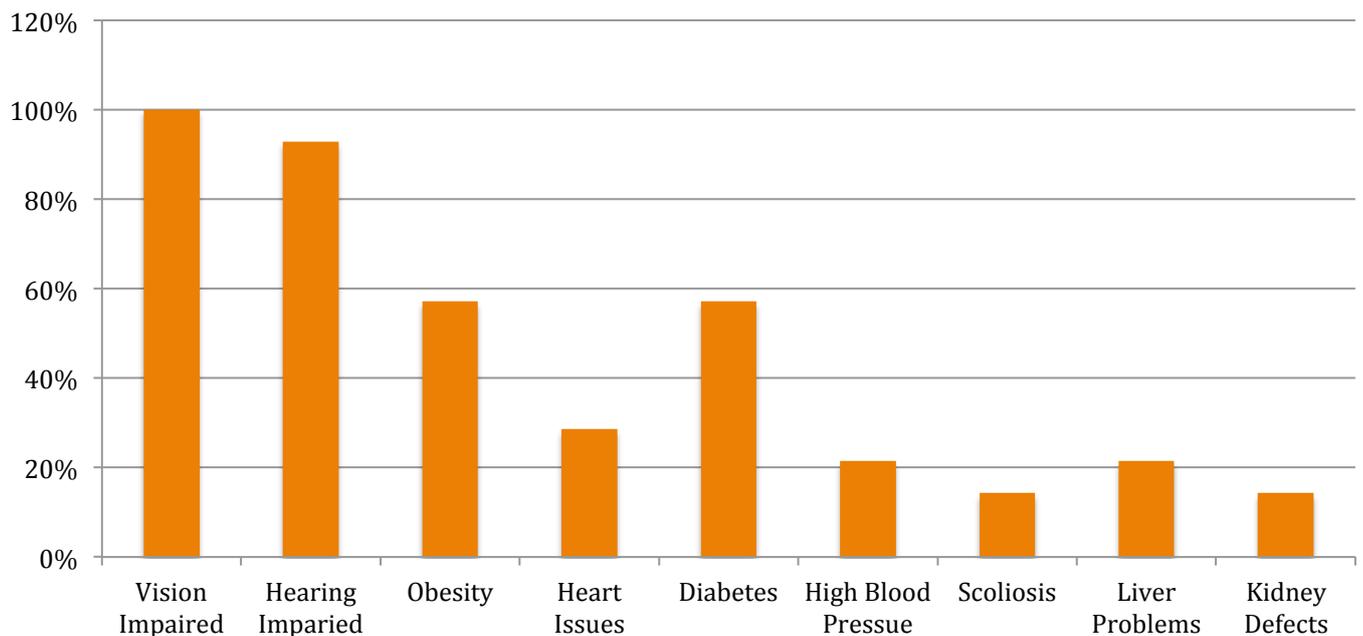


Figure 5 Summary of respondents' symptoms

UK respondents' symptoms in relation to the Italian sample

- On average, the Italian population was diagnosed aged 12, with half the respondents diagnosed under 5, making diagnosis in Italy marginally earlier than in the UK
- Vision loss had occurred in all Italian respondents, as was the case with the UK sample.
- Hearing loss, by contrast, was less common in Italy (in only 14 out of 22 cases) as opposed to 13 out of 14 for the UK
- Heart failure was also less common in the UK with a rate of 4 out of 14, as opposed to 15 out of 22 for Italy

Theme: Collaboration amongst physicians and awareness of Alstrom Syndrome diagnosis criteria

Due to a lack of awareness of Alstrom Syndrome as well as poor collaboration among specialists, there are large gaps in the timely diagnosis of the syndrome and prescription of adequate treatments. Alarmingly, there are also overlaps in medication targeted at different conditions, which have led to negative (and unnecessary) side effects of other organ systems.

According to our survey, we identified the following results:

57%

of patients that reported their diagnosis was as a result of their own personal investigations into the possible cause of symptoms.

54%

of patients that received conflicting information from different health care professionals about treatment options

100%

of patients reported having provided healthcare professionals with information on Alstrom Syndrome

Patients reported that a lack of communication and information sharing between medical physicians caused the following: %

Repeated/duplicated treatments/evaluations/tests	73%
Incorrect prescription of medication	45%
Inefficiencies and timewasting	82%

Theme: Perceptions of networking and information sharing

Currently, the biggest barrier facing research into Alstrom Syndrome is the lack of networking and information sharing opportunities. Without a sizeable sample of patients, it is not possible to perform clinical trials, which inhibits the development of feasible cures. This is particularly concerning given that research into Alstrom Syndrome has the potential to not only develop cures for Alstrom patients but also for single symptoms of Alstrom Syndrome that may affect a far larger proportion of the population. With Alstrom Syndrome's symptoms representing 5 "priority diseases" identified by the World Health Organisation, (WHO) research and developments have widespread ramifications for the whole of Europe.

According to our survey we identified the following: %

Patients agreed that there aren't enough opportunities to access support structures and other families suffering from Alstrom Syndrome	77%
Patients thought physicians and researchers did not have enough opportunities to network with other healthcare professionals to learn how to treat the syndrome	85%
Experienced situations whereby patient information was not openly shared, slowing down the progression of treating Alstrom Syndrome and its future prospects	54%

Key insights into the themes of diagnosis delays, collaboration among physicians and the importance of information sharing

- One problem is that GPs do not jointly treat or interlink all the symptoms (specialists often neglect interdependencies between different symptoms' treatments and medication)
- Often diagnosis is made "by chance", not by systematic analysis
- Lack of awareness of AS along physicians makes diagnosis hard
- Patients do research themselves and frequently self-diagnose themselves
- Early diagnosis is important because it reduces uncertainty and because education support for patients and carers (e.g. IT skills for blindness) can be provided
- Early diagnosis enables physicians to offer genetic counselling earlier

Theme: Economic impact of diagnosing and managing Alstrom Syndrome

The economic implication of diagnosing and managing illnesses such as Alstrom Syndrome are significant and ultimately come at a steep price. The long road for Alstrom patients frequently includes numerous tests and physician visits

According to our survey, we identified the following results:

79% of Alstrom Syndrome sufferers that received additional funding from Social Services by way of direct payments, in assistance of the care for the patient with Alstrom Syndrome

Patients experienced the following in comparison to other more common illnesses, as a result of Alstrom Syndrome

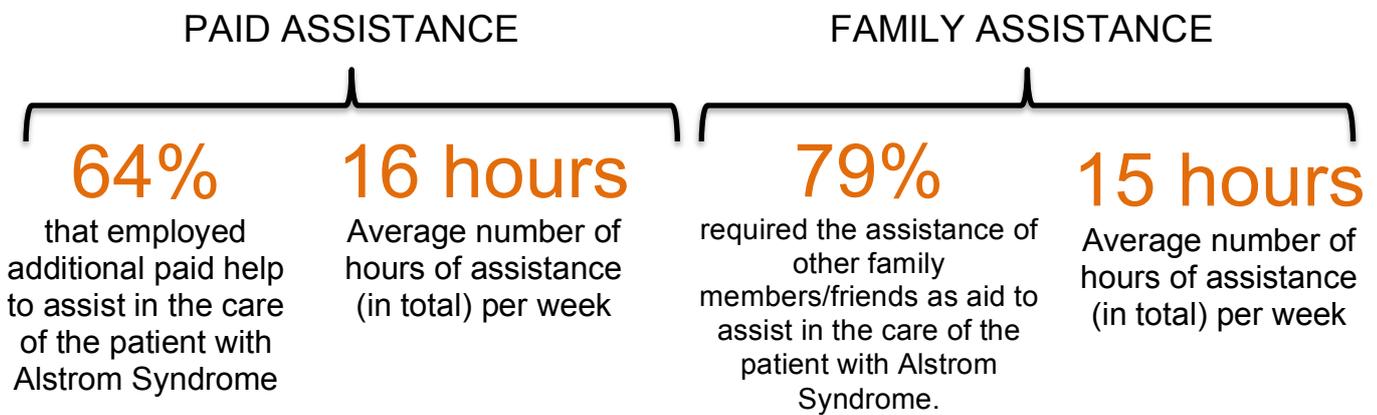
More diagnosis tests	75%
More complex, detailed and costly tests	67%
More visits to specialists	83%
More mental health support	33%

Figure 6 Patients' experience with AS in relation to common diseases

Theme: Financial Consequences of diagnosing and managing Alstrom Syndrome

Managing Alstrom syndrome effects many different facets of life, including families' financial security. Handling the financial aspects of Alstrom Syndrome, which can be exacerbated by bills for special care, travel to find specialists and the inability to work, can become overwhelming.

According to our survey we identified the following results:



According to our survey we identified the following:

	%
Acquired private medical cover to assist financial with the management of Alstrom Syndrome	7%
Used savings to pay for medical expenses	40%
Incurred other direct medical expenses not covered/provided by the NHS	20%
Borrowed additional money to pay for medical related expenses	20%
Sought help from charities or have received public assistance	80%
Alstrom Syndrome medical expenses caused a negative impact on their credit score	10%
Used retirement funds to pay for Alstrom Syndrome expenses	10%
Turned down promotion/business opportunities as a consequences of managing Alstrom Syndrome	40%
Patients that were not eligible to claim travel expenses from the NHS	57%

Figure 7 Qualitative survey results

Percentage of patients that reported incurring additional costs in relation to:

- 92% - Specialist equipment
- 23% - Special needs schooling
- 15% - Psychological therapy
- 62% - Caregivers
- 69% - Medical Supplies (Glasses, hearing aids, incontinence pads etc.)
- 15% - Employed assistance to accompany patients during visits to clinics/physicians

Theme: Emotional consequences and the perceived health-related quality of life

Alstrom Syndrome has a considerable emotional impact on patients, their families and caregivers. The results produced by our health-related quality of life measure resulted in significantly lower results for Alstrom patients compared to participants who were otherwise healthy.

Patients – Living with Alstrom Syndrome Caused	%
Feelings of depression	83%
Feelings of anxiety/stress	100%
Less interaction with family/friends	67%
Worry about how health will change in the future	75%
Worry caused by lack of information on the condition	42%
Them to feel that they had no one to turn to in the medical system for information and support	42%

Figure 8 Psychological factors for AS patients

49.6%

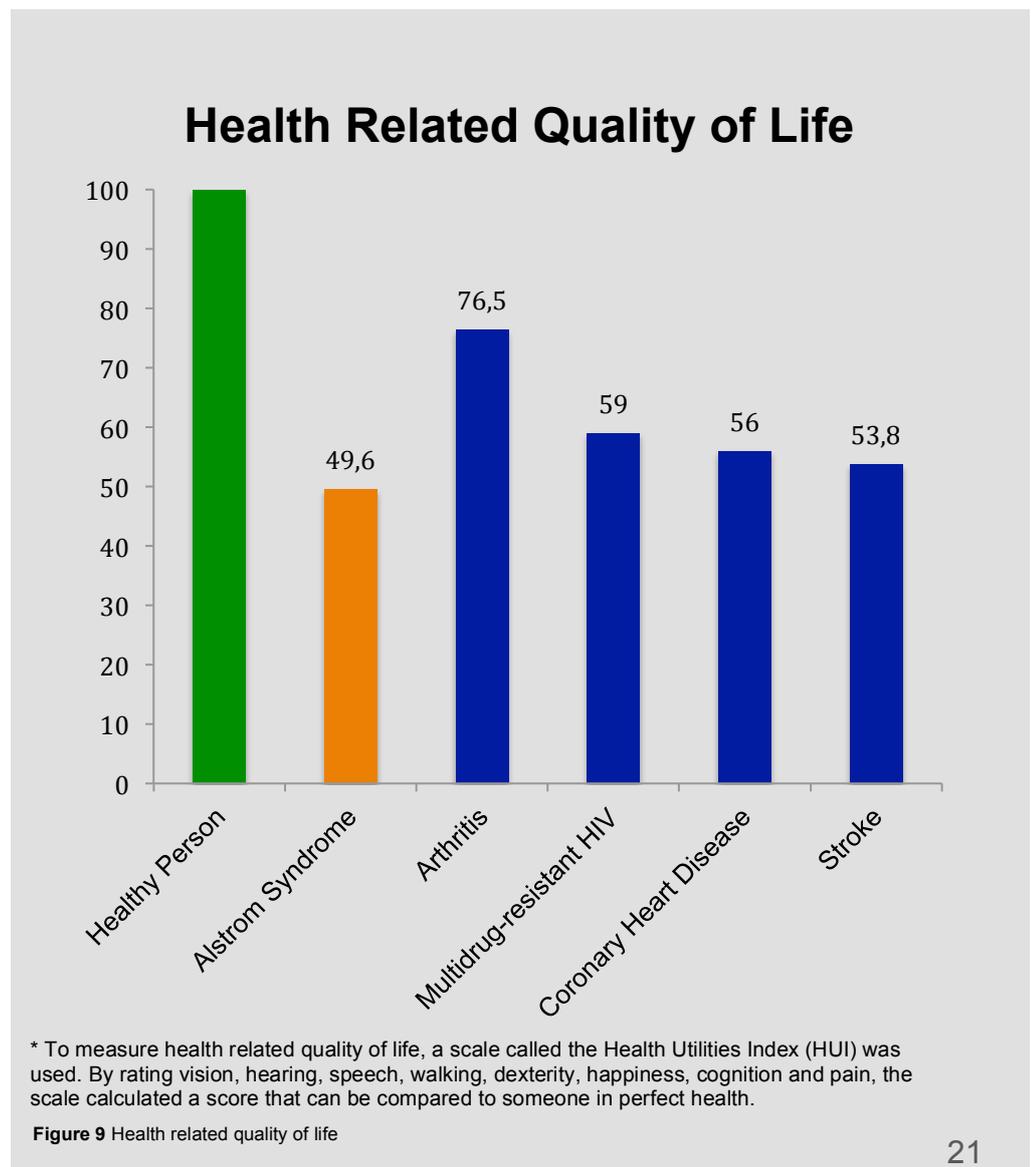
According to the survey results, health related quality of life for patients with Alstrom Syndrome is estimated to be about 49.6% of what it would be if the patients were health

14%

of patients received psychological therapy before the establishment of ASUK's multidisciplinary clinics.

29%

of patients that received psychological therapy (out side of the ASU's multidisciplinary clinics) after the establishment of ASUK's



Caregiver – Caring for Alstrom Syndrome Patient Caused %

Feelings of depression	42%
Feelings of anxiety/stress	83%
Less interaction with family/friends	50%
Worry about how health of the patient will change in the future	100%
Worry caused by lack of information on the condition	42%
Them to feel that they had no one to turn to in the medical system for information and support	50%

Figure 10 Psychological factors for AS carers

8%

of caregivers that received psychological therapy before the establishment of ASUK's multidisciplinary clinics

17%

of caregivers that received psychological therapy (out side of the ASU's multidisciplinary clinics) after the establishment of ASUK's multidisciplinary clinics

92%

of caregivers felt that they had taken on multiple roles, as for example a care co-ordinator, research and advocate, in an effort to receive optimal care and improve the future prospects of the disease

Researcher

Care Coordinator

Advocate

Feedback on the impacts on family life as a consequence of living with an Alstrom Syndrome patient:

Impact on siblings and other members of the family

"Two step siblings got very jealous of the amount of time we had to spend on hospital appointments etc., this has led to a complete breakdown in communication with them and us"

"A very difficult time when first diagnosed - we try to remain positive. Our focus is on ensuring [...] has a excellent quality of life full of opportunities"

"Her brothers are very concerned if they carry the same gene also as they are coming up to an age now to be settling down and starting families."

"Neglect of one child over the other- when a child is ill -this is all you can focus on no effect"

"Devastating!"

" The condition and the associated disabilities have changed the whole family structure. My wife is now working full time and is the only income earner, my son has seen his father become a changed person having to adapt to his now way of life. His daughter was very young at diagnosis so her recollection of her dad is the way he lives day to day now. She may be the youngest but she is the one who supports him the most "

Impact on marital relationship

"Very strained and no time for each other-only time for the children"

"It has been a very difficult time for all of us - the pressure mounts up from time to time"

"As a father it has been difficult to join in many dad things. He goes to his daughters dance shows but has never seen her dance if that makes sense. He has watched his son learn to drive but couldn't offer to go out with him until he had passed his test. Simple dad things that he would of looked forward too but just can't."

"I have the gene, I am devastated that I have somehow passed this on to my daughter and caused her great pain and suffering. It is very difficult to come to terms with as a parent as I have always protected my children 100% and I feel I have let them all down"

Key insights into the Economic Impacts of Alstrom Syndrome

Economic impact of Alstrom Syndrome can be divided into different themes:

Economic impact arising from patient:

- Since the way the patient can contribute to and participate in the economy, is influenced by AS one important economic factor is the reduction of employability of the patient
- The fact that some of the patients study and had good career perspectives before the diagnosis, shows that there is a lot of talent and potential lost

Economic impact arising from caregivers:

- Since family members and other caregivers invest time and effort for the care of the patient, from an economic perspective, there is also an impact
- Caregiver may give up their fulltime jobs or reduce the work time which has an economic impact

Key insights into the Financial Impacts of Alstrom Syndrome

- As mentioned in several parts of this report the financial impact of the Alstrom Syndrome on the patients and their families stems from several issues
- Of course there is a financial impact caused by medication that has to be provided; however, these costs are carried by the NHS
- But there are many indirect costs that the families face
 - Infrastructure/facilities: house of parents has to be adjusted to needs of blind people
 - Education: schools for blind people require different learning material
 - Financial impacts that follow from the economic impacts and the reduced career perspectives described above: families/ patients will have less disposable income

Key insights into the Emotional Impacts of Alstrom Syndrome

- Patients feel like they depend on others
- The diagnosis is very depressing at first, but then patients learn to live with it
- Feeling of isolation often goes hand in hand with a diagnosis of AS: Community of ASUK helps to stop patients from being isolated
- Patient see ASUK as their extended family because people have similar experience which they can share
- ASUK represents a strengthening environment, and one where patients can freely talk about their experiences and worries.

Theme: Impacts of eHealth Solutions

The World Health Organisation defines eHealth as the "use of information and communications technologies in support of health and health-related field, including health care services, health surveillance, health literature, and health education, knowledge and research". eHealth has the potential to completely transform the way Alstrom Syndrome is managed. Using our survey we obtained a better understanding of patient and caregiver perceptions toward proposed solutions.

According to our survey we identified the following results:



Figure 11 Survey results on impact of eHealth

eHealth technologies are able to gather different forms of information on a continuous and real time basis.

Patients felt the following platforms would provide the best access to electronic health records of patients living with Alstrom Syndrome

Patients felt the following types of information were the most useful in relation to managing Alstrom Syndrome?

Mobile phone	36%	Diet	100%
Tablet	64%	Exercise vitals	79%
Laptops	64%	Heart rate	71%
Desktop PC	64%	Blood sugar level	86%
Other: Face to face (7%)		Blood pressure	86%
		Other: Body temperature (50%), Blood Tests (7%)	

Figure 12 Patients' evaluation of eHealth services

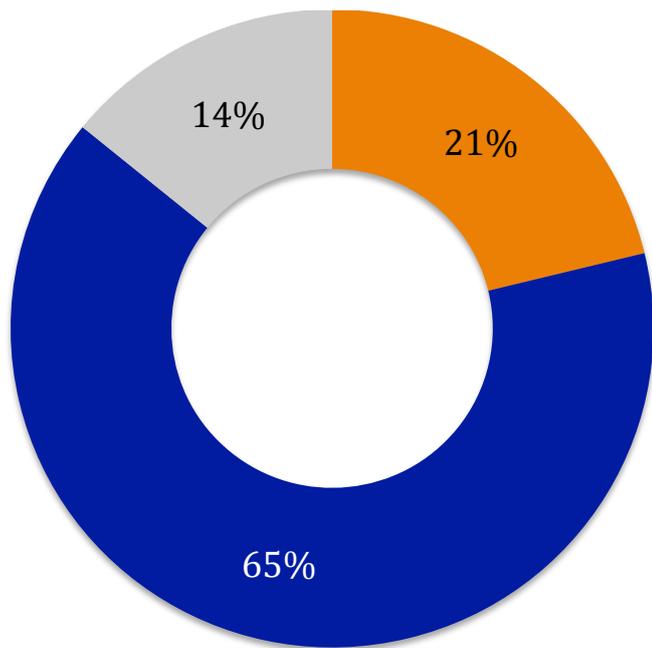
Patients felt they would use eHealth solutions to monitor their health

Ranking of services in order of importance to patients

Daily	43%	Insulin and sugar reading devices	1 st
2-3 times a week	21%	Blood pressure monitor	2 nd
Once a week	14%	Weighing Tools	3 rd
Once a month	1%	Pacemaker monitor	4 th

Figure 13 Patients' evaluation of eHealth services (cont.)

Theme: Impacts of eHealth Solutions (continued)



% of patients felt comfortable with having a virtual relationship with physicians and specialists that may not be physically present during diagnosis and consultations?

- Yes, regardless of whether it replaces face to face consultations
- Yes, only if you felt assured it would not replace face to face consultations
- Not comfortable

Figure 14 Patients' acceptance of eHealth

79%

of patients felt that applications monitoring their cardiac rhythm and insulin levels would provide them with reassurance

57%

of patients felt that applications monitoring their cardiac rhythm and insulin levels would reduce the amount of consultations they were required to attend and their overall travel time

100%

of patients felt comfortable having their medical information on a "cloud" software making it accessible by specialists in any location (e.g. in other countries), in order to stimulate better coordination and research capabilities?

Respondent sample had the following concerns regarding the use of eHealth solutions

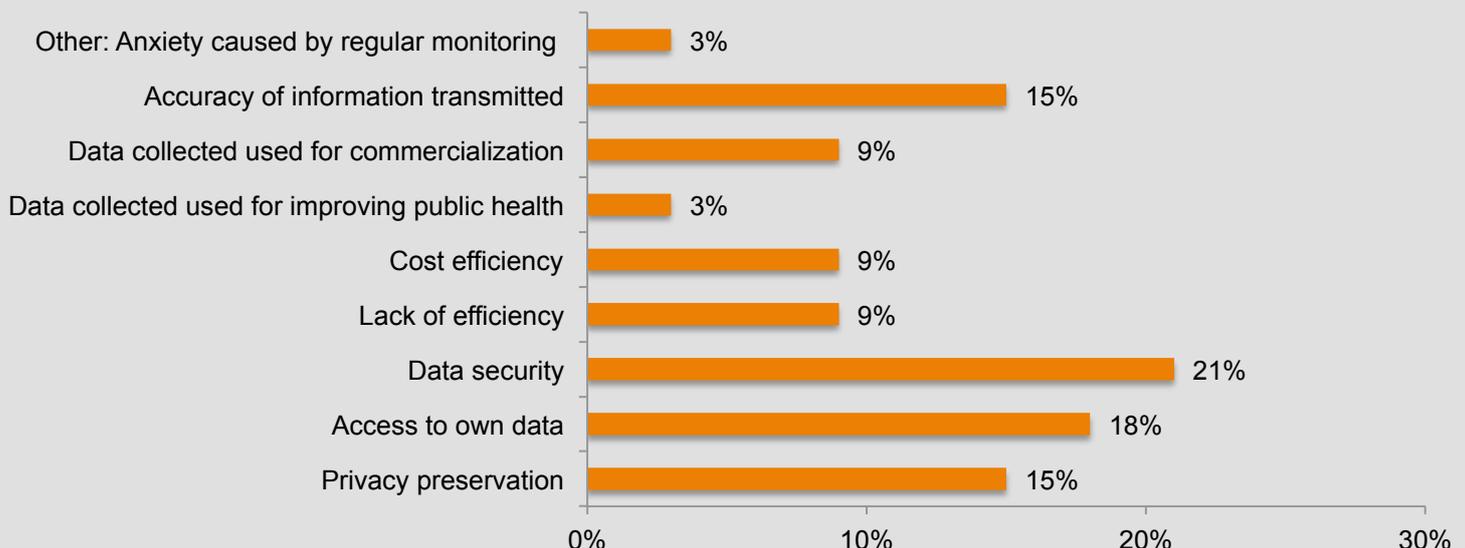


Figure 15 Concerns regarding eHealth

Key insights into proposed eHealth solutions

Physicians' perspective on eHealth:

- eHealth application can help to reduce patient anxiety and also reduce the number of treatments that the patients have to undergo outside the MDC.
- According to the physicians, eHealth would not replace MD clinics, but would rather serve as an additional service for patients and reduce the number of visits to their local GP.
- Patients would not be “reliant on an ignorant local doctor” but could contact their specialist via an eHealth application.
- For example, blood sugar levels could be measured and insulin injections adapted to short term patient needs (i.e. blood sugar levels). The patients could even receive advice from doctors over the eHealth application.
- Data protection and regulation may be an issue: Ownership of eHealth data is not fully resolved, as there are three parties involved:
 - The hospital or patient that uploads data,
 - The service provider (e.g. Orange) who transmits the data, and
 - The hospital or doctor that downloads the data

➔ Clearer regulation is therefore required to establish rules and norms (especially in cross border cases!)
- Also the format of the data that is shared needs to be standardized within the EU.

Patients' perspective:

- Patients see the potential of eHealth in improving accessibility and ease of contact.
- It is crucial that the technology is tailored to AS patients: especially the accessibility of the gadgets is really important due to dual sensory loss.
- Some patients already use tablets and are able to send and read emails
- Often, at clinics, patients might say “I had this problem 3 weeks ago” – this makes treatment difficult and often the condition might have become worse.

➔ It would be a major step forwards if patients could simply send an email or a device would monitor and send doctors live updates.

Theme: Morality Costs

Methodology: Costs attributable to premature mortality are calculated as the current value of lifetime earnings lost according to age. For our Alstrom Syndrome calculation we created a distribution of deaths by age from our survey results, research and access to medical databases. The sample was then used to extrapolate the results for the U.K. Alstrom Syndrome population as well as the worldwide Alstrom Syndrome populations. This same approach was used in calculating morality costs for rare diseases. However, given that over 7000 rare diseases have been identified globally, we used a normal distribution of deaths by age to ensure a more realistic representation of the population. Earnings lost are discounted to the present at rates reflecting historic GDP growth and inflation.

£ 20 281 392.14

UK Alstrom Syndrome Life Time Earnings Lost

\$ 1 770 208 549.76

Worldwide Alstrom Syndrome Life Time Earnings Lost

£ 1 179 793 169 164.29

UK Rare Disease Life Time Earnings Lost

\$ 85 574 939 736 287.70

Worldwide Rare Disease Life Time Earnings Lost

Interpretation of Results

Results of the mortality cost calculations should not be interpreted as annual amounts. Instead, they represent the earnings that would be lost over a lifetime, due to premature mortality caused by Alstrom Syndrome and Rare Diseases. The major assumption made is that these populations will have the same distribution of deaths according to age as used in the calculation. Results should thus be interpreted as the lifetime earnings lost by the populations used, assuming they follow the same distribution of deaths according to age.

Key Assumptions

Working Age:	18 – 60 Years
Average Earning (UK):	GB£24 816 p.a.
Average Earnings (Worldwide):	US\$18 000 p.a.
Prevalence of Alstrom Syndrome:	1 : 1 000 000
Size of Alstrom Syndrome Population (UK):	60
Size of Alstrom Syndrome Population (Worldwide):	7 220
Size of Rare Disease Population (UK):	3 500 000
Size of Rare Disease Population (Worldwide):	3 500 000 000
Mean Worldwide Age:	28.85 Years
Standard Deviation (Worldwide Age):	8.50 Years
Discount Rate:	6%

Key insights into costs associated with premature mortality

- Premature mortality of Alstrom patients accounts for a life time earnings loss of more than GBP 20mil only in the UK
- Worldwide premature mortality of Alstrom patients accounts for a lifetime earnings loss of more than USD 1.7bn .
- Numbers show the huge impact of the Alstrom syndrome and underline the importance of research in this rare disease
- Considering rare diseases in general, the numbers are even more impressive and show the importance of research
- One also has to keep in mind that research into rare diseases could possibly even have an impact on non-rare diseases and therefore have even bigger impact on Earnings Lost

Conclusion

Based on the survey results presented above, there are a number of core conclusions that can be drawn.

First and foremost, the value of the MDCs to patients is extensive and far-reaching. Patients praised the expertise of the doctors, time savings, easy access to treatment and planning security. It is true that there are also some weak points to the centralised clinics, such as the effort required to coordinate them (from practitioners' perspective) and the travel distance (from patients' perspective), yet all factors considered both patients and health care professionals agreed that the advantages far outweighed the costs.

What is more, the value of the clinics can be seen to far exceed the value of each individual treatment; in this sense, the MDCs have turned into a place of exchange for AS patients and specialists and provide real support for families in need. Any cost benefit analysis of the clinics therefore needs to look beyond the pure treatment benefits to also include this psychological support function of the clinics. This is apparent in both the caregivers' and patients' responses.

Finally, from a societal perspective, there is both a moral argument to reduce morbidity costs of AS patients and also an economic argument relating to lost earnings resulting both from the condition itself and the time lost to treat it. This affects both the patient and the caregiver, and more effective, time efficient treatment practices arguably reduce the burden on society from lost days' work. Furthermore, the following recommendations can be made:

- **Diagnosis:** Continued efforts should be invested into diagnosing AS as early as possible. This could be achieved by raising awareness for the condition among physicians or through improved predictive technology and family counselling in high risk cases
- **Treatment:** Due to GPs' and local hospitals' lack of knowledge on the condition, closer integration between specialists and patients needs to be achieved. One way to achieve this is by employing eHealth technology
- **eHealth applications:** The patients' concerns regarding the accessibility of eHealth apps (as revealed in the survey and interviews) need to be addressed, essentially by service providers.

The Italian Population

Objectives

By working together with Dr Pietro Maffei and Dr Vero Buttini from EUROWABB, we were able to gain access to medical statistics obtained from 22 Alstrom patients in Italy. Our objective in so doing was to create a benchmark and reference point for our UK results from the patient survey, and to thus have a more founded idea of whether our survey responses were representative of a broader cohort of Alstrom patients.

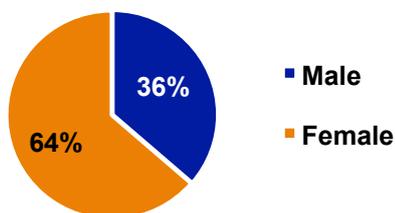
This approach is particularly salient for three main reasons:

1. First, it is a core measure to counter the concerns regarding sample size and selection bias. These result mainly from Alstrom's nature as an ultra-RD.
2. Second, one of the objectives of this research report is to establish a European vision for AS treatment. It is therefore highly relevant to gain insights from more than one member state. In further research, the sample of Alstrom patients should be further extended to gain an even more accurate picture of the condition and its related symptoms across the EU.
3. Third, cooperation between research bodies is one of the core recommendations not only of this report, but many others beside it. International exchange of medical data is therefore a first step to developing best practices in treatment and collaborative cross-border research.

Population fundamentals

4:7

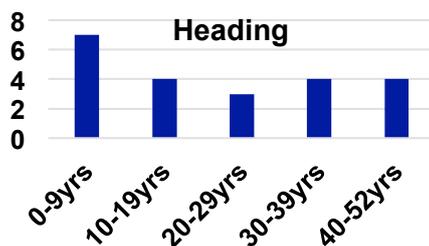
Average ratio of male to female patients in the sample



The vast majority (nearly two thirds) of the sample was found to be female. This stands in contradiction to Marshall (2005) where only 48% (i.e. less than half) were female.

22

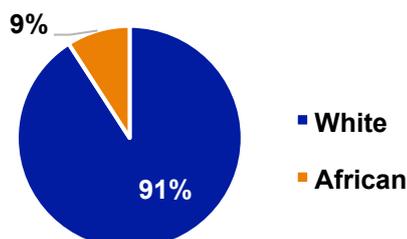
Average age of patients in the sample



The average Italian age was much in line with the British sample. But almost surprisingly, the Italian sample was nearly exclusively white, which stands in contrast to the Asian families found in the UK.

90.9%

Of patients in sample is ethnically white



The ethnicity of the Italian patients was an interesting point, as in the UK there has been a higher prevalence of Alstrom among some ethnic groups in the UK (e.g. Asians). This is due to cultural factors.

Figure 16 Italian population fundamentals

Symptom related data

Diagnosis

It is evident that diagnosis of Alstrom occurs primarily in the early childhood of the patient; it should be noted that the average age of the first group (0-4yrs) is 1.7, indicating that a large number of AS sufferers is diagnosed within the first two years after birth. What is more, our qualitative investigation revealed that diagnosis seems to be occurring at an ever-earlier stage, which may bring some treatment benefits. However, there also seems to be a small group of Alstrom carriers that are only diagnosed quite late in life, i.e. 30 and above. Further medical research would be warranted into this (seemingly) separate “late-developing” patient group. It should also be added that 4 of the patients below are included in the sample with their age at death, which was 3months, 23yrs. 30yrs and 48 years respectively.

Dispersion of age diagnosed

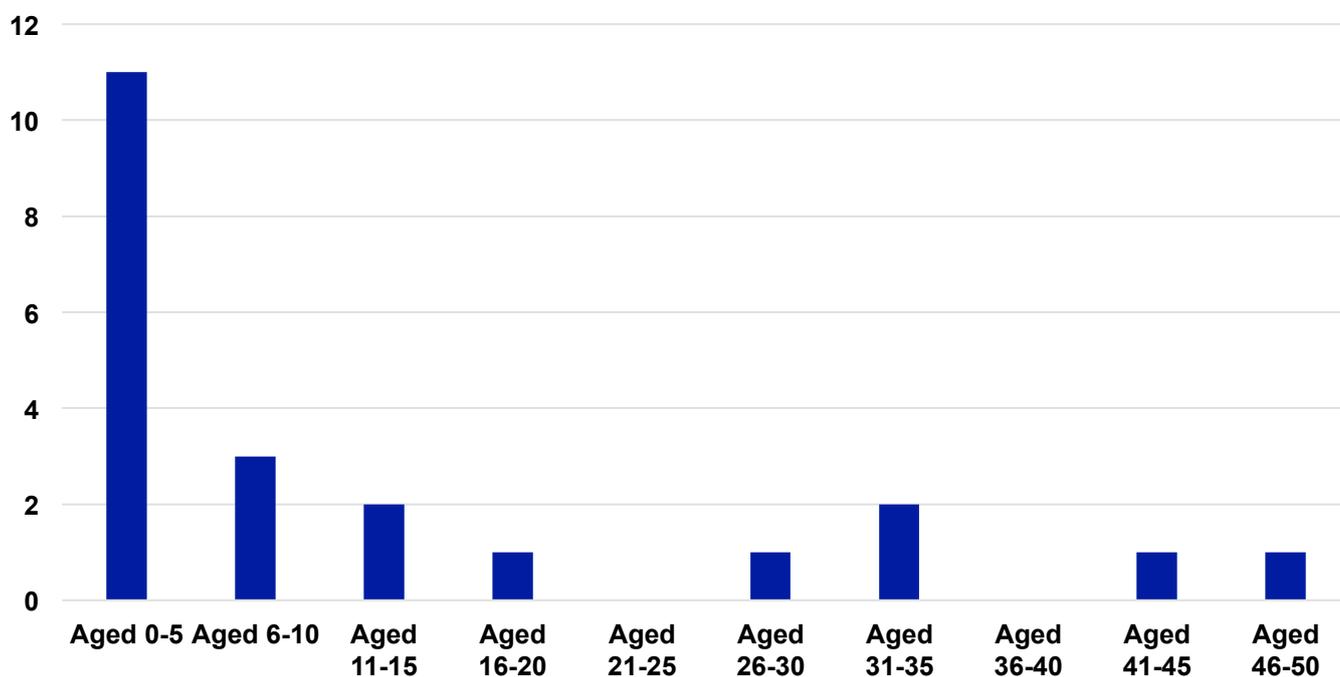


Figure 17 Italian dispersion of age diagnosed

Symptoms exhibited

Thanks to the very detailed work of the Italian scientists, a detailed picture of the Italian population’s symptoms is presented below. A score of 22 thereby means that every patient surveyed exhibited this symptom.

3	3.86	8.91
Number of patients with symptoms in all six major categories	Average number of categorised symptoms per patient	Average number of symptoms per patient

Symptoms exhibited by the Italian patients

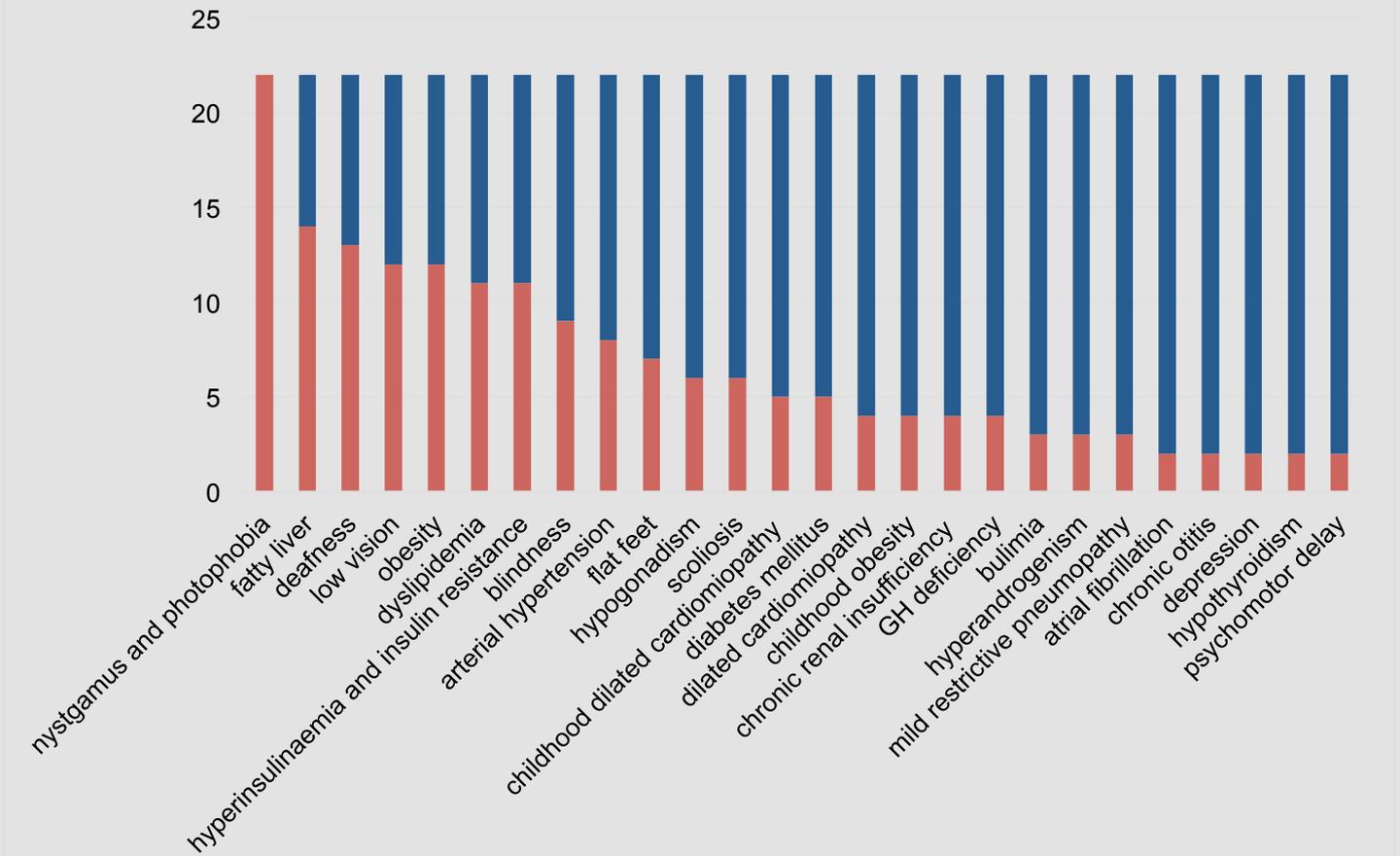


Figure 18 Italian detailed overview of symptoms

In addition to the major symptoms included in the graph above, the following rare symptoms were listed (by a maximum of 1 patient each)

- Bipolar disorder
- Brain ischemia
- Bronchial asthma
- Flat feet
- Frequent bronchitis and bronchial spasm
- Heart insufficiency
- Hypothyroidism
- Impaired glucose tolerance
- Kyphoscoliosis
- Liver cirrhosis
- Microcephaly
- Muscular hypertonia
- Nocturnal enuresis
- Nutritional problems
- Osteoporosis
- Psoriasis
- Psychological problems
- Pulmonary hypertension
- Recurrent otitis
- Restrictive pulmonary disease
- Urinary and faecal incontinence

In order to ensure comparability of results between the Italian and the UK patients, the symptoms were classified into six categories: vision, hearing, obesity, heart, kidney and liver. The results are displayed in the graph below.

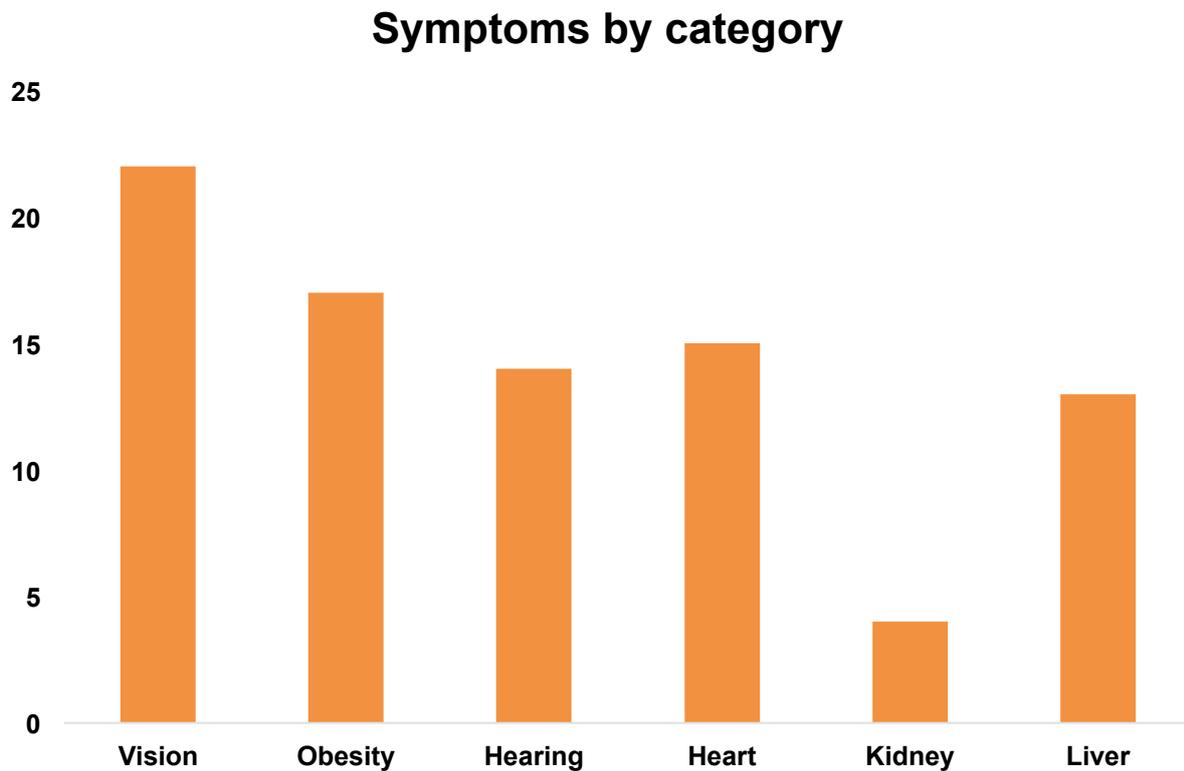


Figure 19 Italian symptoms by category

It is interesting to note that the average number of (categorised) symptoms exhibited per patient was 3.86. This once again points out the necessity for multi-disciplinary treatment in light of the diverse conditions.

Qualitative input obtained from scientists

In addition to the questions surrounding the patients' respective medical conditions, a series of qualitative questions was asked to gain more insight into the treatment practices in Italy. Data related to these questions was not available for 4 patients; therefore the percentages are out of 18, not 22.

Percentage of patients that...	%
...needed fulltime care	0%
...had access to treatment	100%
...were forced to travel for treatment	100%
...needed to be treated abroad (within the EU)	0%
...complained about prior misdiagnosis of AS	22%
...had comprehensive medical insurance	100%

Figure 20 Italian qualitative results

Review of the benefits of eHealth

eHealth Solutions

Introduction of eHealth

Multi-disciplinary clinics (MDC) have been successful at consolidating both treatment practices and knowledge to fight rare diseases. The dissemination of mobile devices, smartphones, and apps has facilitated healthcare provision and communication. These improvements in healthcare management and technology have provided new opportunities for the integration of mobile health into existing clinics. Therefore, the implementation of a new model of a virtual multi-disciplinary clinic is a likely next step towards better effectiveness and efficiency in the diagnosis, treatment and management of rare diseases in general and AS in particular.

At the heart of these technological and clinical improvements is mobile health (eHealth). To date, no standardised definition of eHealth has been established. According to the World Health Organization (2011), it can be characterized as the “use of mobile and wireless technologies to support the achievement of health objectives”. As a subcomponent of the larger discipline of eHealth¹, mobile health is said to have “the potential to transform the face of health service delivery across the globe”² due to its wider accessibility via mobile devices, smartphones and self-monitoring gadgets. However, the European Commission’s Digital Agenda for Europe stresses, “mobile health doesn’t focus exclusively on the device, but on the fact that the information and data is mobile”.

eHealth technologies provide solutions in relation to personalized health technologies, mobile/portable apps, co-operative ICTs, sensors and devices, software and others for monitoring purposes and providing diagnostics. These technologies enable remote diagnosis, build disease-specific networks, as well as deliver healthcare information/data enabling patients’ self-management. A wide range of stakeholders (patients, health care professionals, researchers, charities, etc.) therefore has the ability to converge and pool data using eHealth technologies to create one integrated platform.

Methodology and Objectives

Although eHealth has become a key emerging tool in the healthcare industry, it lacks adequate research, evidence and sophisticated usage in relation to rare disease management. In this report, we will analyse the benefits and drawbacks of eHealth, the different providers and eHealth platforms currently available to ASUK, and examine the potential utility of these platforms for other rare disease charities. In order to deliver this analysis, we have reached out to healthcare professionals, patients, academic studies, product solution providers as well as charities using methods including a literature review, desk research, interviews and surveys. Through this research, we aim to show the following:

- Show benefits, drawbacks and concerns of eHealth applications and their potential applicability to AS patients
- Shortlist eHealth providers and make a recommendation regarding a strategic partner for ASUK
- Determine how ASUK’s business model could be replicated by other URD charities

¹ eHealth, refers to the use of Information and Communication Technologies (ICT) for health purpose

² WHO (2011), eHealth. New horizons for health through mobile technologies

Potential Benefits & Disadvantages of eHealth and its relevance to ASUK

We examine the benefits, drawbacks and concerns of eHealth based on an extensive literature review and based on interviews with healthcare professionals and patients. Although eHealth provides benefits to patients, patients' families and healthcare providers, it also has several limitations, such as concerns about adopting eHealth for broad application in healthcare practices. Bearing these concerns in mind, we analyse the ways to capitalize on the benefits of eHealth while controlling the risk and managing those concerns.

Benefits of eHealth

We will consider the benefits with regards to patients and to healthcare service providers.

Benefits to patients

eHealth provides the possibility to integrate multi-disciplinary clinics with the help of an accessible platform via mobile virtual clinics. Ideally, patients and doctors would be able to attend MDCs without being obliged to travel and or stay abroad for several days. Instead, they would be required to log onto the eHealth platform and could communicate online with each other³.

A second benefit of eHealth and the community platform would be that patients and families receive services, disease management information and education online. This community platform would therefore save a variety of costs for patients and their families, such as seeking emergency help for non-emergencies, having multiple specialist consultations and other opportunity costs like work absenteeism due to hospital consultations. A study conducted by The Department of Economics at University of Gothenburg evaluated the benefits of the Ågrenska Centre in Sweden; a healthcare centre that brings children with rare disorders and their families together with support services. The study shows that the holistic approach taken by Ågrenska Centre has the potential to cut the average costs for treatment for a child from 35,000 Swedish kronor (SEK) to 13,000 SEK per year⁴. This amounts to possible savings of \$US350 million annually for Sweden. For Europe, the savings could be as high as \$US10 billion annually⁵. EHealth has the potential to provide the same services as the Ågrenska Centre but at an even lower cost. Therefore, eHealth can potentially contribute to significant cost savings for patients and their families.

Thirdly, eHealth has been shown to improve the quality of healthcare services by enabling early identification of potential diseases, providing devices for self-management, improving communication between patients and doctors, and delivering a response in emergency situations. Using eHealth it is possible to collect medical data from patients such as their body temperature, blood pressure level and heart rate via wearable devices. These wearable devices act as sensors that measure and collect health information and update it onto the cloud-computing database. Both patients and doctors have access to this data, and doctors and nurses can check the real-time health condition of their patients. Once any symptom is noticed, doctors can contact patients immediately and advise them on the necessary actions.

Furthermore, eHealth enables patients to manage their symptoms by themselves and communicate with doctors easily and conveniently. Patients receive constant advice from doctors and follow-up reminders to take their daily medication. They also have access to their electronic health record (EHR) to track their own health condition on regular basis. In addition, relevant doctors and hospitals are alerted immediately in emergency situations when sensors would detect

³ Interview with Prof. Tim Barrett from Birmingham Children's Hospital

⁴ *The Ågrenska Centre: A Socioeconomic Case Study of Rare Diseases*, Anders Olauson, Ågrenska Centre for Rare Disorders, Gothenburg, Sweden

⁵ *The Ågrenska Centre: A Socioeconomic Case Study of Rare Diseases*, Anders Olauson, Ågrenska Centre for Rare Disorders, Gothenburg, Sweden

patients' unusual or alarming symptoms. This allows for a quick reaction and solution to emergencies that at times could be fatal if they were not quickly dealt with.

Fourthly, eHealth improves the patients' quality of life. With eHealth, patients are able to reduce their hospital and clinic consultations by receiving remote healthcare services at home. Information communication technology provides patients with an easy and convenient way to get information about their own health conditions, to get advice from healthcare professionals and to communicate with people with same diseases.

Benefits to healthcare providers

First and foremost, eHealth has the ability to improve the efficiency of healthcare providers. Healthcare providers can automate healthcare administration services, streamline administrative documents and back-end processes related to the provision of healthcare. Therefore, eHealth can have a positive impact on the efficiency of the overall healthcare service delivery system. For example, an appointment reminder function can reduce non-attendance rates and also improve patients' experience of the outpatient care process.

Moreover, eHealth improves the communication among healthcare professionals. In one meta-analysis conducted by Free C, Phillips G, Watson L, Galli L, Felix L, et al, the use of eHealth contributed to improvements in nurse and surgeon communication⁶. EHealth also helps healthcare providers to improve their operation efficiency. In a study, Pearson reported a revenue increase of 5% to 20% when doctors billed patients on the move using Patient Keeper software on Personal Digital Assistants (PDAs) at the University of Kansas Medical Centre⁷.

In addition, eHealth provides healthcare practitioners with support including mobile access to IT systems and sophisticated databases– from the simple look-up of information (e.g. medical encyclopaedias) to intelligent decision support systems that aid in the diagnosis and treatment of patients. This feature also includes the dissemination of medical information, training, and updates relevant to healthcare practitioners.

Drawbacks and Concerns of eHealth

The main drawbacks of and concerns around eHealth regard the effectiveness of healthcare services delivered via eHealth and data related issues.

Effectiveness of healthcare services delivered via eHealth

The widest concern regarding eHealth is the effectiveness of healthcare services delivered via eHealth. Many patients and healthcare professionals are reluctant to adopt a broader application of eHealth due to concerns of misdiagnosis. Dr. Tarek Hiwot from University Hospitals Birmingham (UHB) mentioned that according to his experience, the correct rate of self-diagnosis via eHealth is less than 25%⁸. Although this number is based on his personal experience and there is no data from systematic research, it shows an important limitation of eHealth systems. This drawback of self-diagnosis accuracy via eHealth can be due to reasons such as poor connection quality, distortion of pictures sent via the internet, patients' lack of medical knowledge and inadequate monitoring accuracy.

⁶ Free C, Phillips G, Watson L, Galli L, Felix L, et al. (2013) *The Effectiveness of Mobile-Health Technologies to Improve Health Care Service Delivery Processes: A Systematic Review and Meta-Analysis*. PLoS Med 10(1): e1001363. doi:10.1371/journal.pmed.1001363

⁷ H.Pearson, *Doctors Going Wireless with Souped-up PDAs*, 2005, <http://www.msnbc.msn.com/id/710981013/> CNBC News

⁸ Interview with Dr. Tarek Hiwot from University Hospitals Birmingham

This is confirmed in a study conducted by Free et al., which also shows that statistically there are significant reductions in correct diagnoses using mobile technology photos compared to the “gold standard”⁹.

Lastly, it is clear that for some check-ups such as CT scans, patients still need to go to the hospital, which in these cases erodes the convenience of using eHealth.

Data issues

Another major concern of eHealth regards the collection, storage and protection of patient data.

Firstly, the integrity of the online data cannot always be guaranteed. Data is inputted by patients and doctors, stored in a database, and then extracted from the database when necessary. This means that there are three parties involved in the data storage and usage process, making it difficult to prove wrong-doing should any single data point be faulty. According to the interviews conducted, this is particularly relevant in cross-border transactions where there is an insufficient regulatory framework in place.

Moreover, it is difficult to monitor whether the authorized patients or doctors would be able to still have easy access to the data if sufficient security mechanisms are put in place.

Thirdly, as expressed by many patients and the healthcare professionals we interviewed, privacy is a major concern. To capitalize on the benefits of eHealth, it is inevitable that some patient data would be exploited for treatment or research purposes. However, most medical data is highly sensitive, and therefore protecting patients' privacy is critical.

Last but not least, cross border data transfer is a debatable issue in the EU and the application of eHealth must be subject to EU legislation about cross border data transfer. This relates to the need for clear regulation referred to above.

Control Risks and Manage Concerns while Capitalizing eHealth Benefits

To control the risks and manage concerns surrounding misdiagnosis, we suggest that ASUK integrates eHealth merely as a self-management tool for the disease, but not in any diagnostic function. This would involve self-monitoring applications and remote consultation platforms for patients, and would play on the two major advantages of eHealth, namely constant monitoring and efficient information exchange between patients and healthcare providers. This, combined with remote IT systems, such as video conferencing and the use of wearable devices used for monitoring (e.g. heart rate monitor), will enable superior patient services and improve early identification of further symptoms and conditions.

Regarding the data issues identified above, it is possible to apply eHealth with features providing solutions to these issues. Building an eHealth system in which every data input is standardized and tracked is a potential solution in solving the issue of data integrity.

1. Data input: Data input needs to be standardised to be compatible with large scale (ideally international) databases. Users could receive additional IT training on how to correctly input data so as to avoid irregularities
2. Data tracking: Regarding the issue of liability, it is important to set up a system which tracks any data modifications – both in terms of new data uploaded and changes made to existing data. Having such a system in place would furthermore be a vital first step to avoiding vague liability relating to the numerous parties involved in eHealth platforms.

⁹ Free C, Phillips G, Watson L, Galli L, Felix L, et al. (2013) *The Effectiveness of Mobile-Health Technologies to Improve Health Care Service Delivery Processes: A Systematic Review and Meta-Analysis*. PLoS Med 10(1): e1001363. doi:10.1371/journal.pmed.1001363

To solve the concern of data security, a potential solution is to outsource data storage and management to a third party that has high international credibility, either a commercial organization or a research centre. Data could be transmitted to and stored at that third party then extracted when requested by authorized users.

The EU legislation provides clear guidance on protection of patients' privacy in the electronic communication sector in cross-border healthcare¹⁰. Outsourcing data storage and management to a credible third party could partly solve this privacy issue. A technical solution would be to adopt a secure and privacy-preserving opportunistic computing (SPOC) framework, which introduces a user-centric two-phase privacy access control to only allow those medical users who have similar symptoms to participate in opportunistic computing therefore to minimize disclosure of privacy of users of eHealth¹¹.

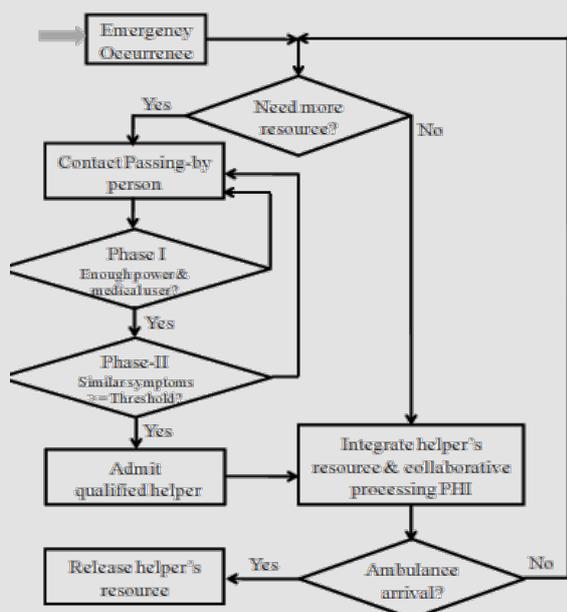


Figure 21 Opportunistic computing with two-phase privacy access control for m-Healthcare emergency

(Source: SPOC: A Secure and Privacy-Preserving Opportunistic Computing Framework for Mobile-Healthcare Emergency)

Conclusion

Based on our analysis, we conclude that it will generate significant benefits and efficiency improvement for both patients and healthcare providers if ASUK adopts eHealth. However, considering the risk of miss-diagnosis and concern of data issues, we suggest that ASUK integrate eHealth as a disease self-management tool, self-monitoring application and remote consultation platform for patients. The adoption of a secure and privacy-preserving opportunistic computing (SPOC) framework can also help to control risk and release concerns.

¹⁰ Directive 2011/24/EU of the European Parliament and of the Council on Application of Patients' Rights in Cross-border Healthcare, 9 March, 2011

¹¹ Rongxing Lu, Xiaodong Lin & Xuemin Shen (2012) SPOC: A Secure and Privacy-Preserving Opportunistic Computing Framework for Mobile-Healthcare Emergency, Parallel and Distributed Systems, IEEE Transactions on (Volume:24 , Issue: 3)

Review of eHealth service providers

Analysis of current eHealth service providers including cost comparison of different partnership models

ASUK has several options of providers for the implementation of a virtual clinic. The choice of the supplier depends on criteria including pricing, services provided (and their adaptability to the symptoms that Alstrom patients can experience), and lastly the availability and easy access across borders for the specialists and the patients. Although this competitor analysis is focused on the specific case of ASUK, it could act as a toolkit model for other rare disease charities and even as a general and aspiring model to implement in hospitals globally. In this section, we will outline the virtual clinic models provided by IBM, Gradient, Cisco, Vodafone and Orange by examining their services, pricing and perceived benefits for patients. After this analysis, we look at a case study that analysed the qualitative benefits of virtual clinics and remote devices.

IBM Healthcare Island¹

IBM has constructed a 3D program called the “virtual healthcare Island” supported within Second Life in 2008. This Healthcare Island provides an interactive dimension of IBM open standard-based health information exchange architecture combining patients and physicians in a virtual world. Through the creation of avatars, members can upload their own personal health records (PHR), implement an updated electronic medical record (EMR) system that can be used by various physicians and medical facilities. Within this software, the avatars can move from island to island depicting different facilities (hospital, laboratory....) with their updated and integrated electronic health records that is safeguarded in a highly secured network. Access to this medical record and PHR is based on a patient-authorized health system allowing only elected family members to view the files. Below are the offer details for IBM’s Healthcare Island:

Offer	3D Virtual Healthcare Island
Platform	<ul style="list-style-type: none">• Patient home• Laboratory & radiology suites• Clinic• Pharmacy• Hospital• Emergency Room
Services	<ul style="list-style-type: none">• Online Personal Health Records• Electronic Medical Record (updated)• Blood and radiology test can be performed• Verification of prescriptions – look at contra indications• Video conferences with specialists
Devices/ Monitors	<ul style="list-style-type: none">• Weighing Tools• Blood Pressure Monitors• Sugar Reading Devices
Pricing	<ul style="list-style-type: none">• No Pricing Communicated

Figure 22 eHealth offers: IBM

¹ <http://www-03.ibm.com/press/us/en/pressrelease/23580.wss> and http://www.medgadget.com/2008/03/ibm_builds_virtual_healthcare_island.html

This system is ideal for ASUK and the patients, as it not only incorporates updated online personal medical records, but also provides a platform for consultations and advice from different specialists. Moreover, patients within their avatar home have the ability to test their blood, their sugar levels and weight without having to consult their local physician saving time, money and costs. Lastly, it allows patients suffering from multiple symptoms, such as those from ASUK, to overcome the multidisciplinary issue by having easily accessible medical records for the various specialists and a pharmacy monitoring the different medical prescriptions.

However this system does present several limitations. Firstly, there is no sign on how IBM and the virtual healthcare island can be used by the blind or patients with limited visual abilities as well as those that experience deafness or hearing deficiencies. These are two primary symptoms of AS and therefore need to be addressed by IBM in order to provide a fully functioning system tailored to dual sensory loss patients.

Secondly, even after having attempted to reach IBM several times, there has not been a response by the corporation in terms of the tailoring of their service and their price offered. This makes it difficult to analyse in Alstrom’s case for cost and economic impact on the charity.

Gradiant²

Gradiant is an acronym for Galician Research and Development in centre in Advanced Telecommunications, which was created as a non-profit private foundation in Spain. Gradiant targets the transfer of knowledge to industry, to take responsibility for the R&D of their clients with their R&D and ISO certifications. This Spanish telecommunications organization is specialized in four technical areas that it can offer to its variety of clients. Although Gradiant is based in Spain, it acts as a viable competitor and supplier of ICT or mobile health knowledge for ASUK to use through their multi-disciplinary model with specialists, physicians and patients.

Here below is a descriptive of Gradiant’s offer:

Offer	Gradiant
Platform	<ul style="list-style-type: none"> No Integrated platform or product
Services	<ul style="list-style-type: none"> Consulting, Prototyping and Advice on: Data Protection Communication Abilities Technical Knowledge Ability of multimodal information
Devices/ Monitors	<ul style="list-style-type: none"> No product but can prototype devices
Pricing	<ul style="list-style-type: none"> Pricing based on consultation/hour (varies depending on seniority of consultant and length of project)

Figure 23 eHealth offers: Gradiant

It is clear that Gradiant is also a competitive option. It specializes in the four technical areas required for effective virtual clinics including digital communication, applications and networks, multimodal information and eHealth.

² Gradiant Corporate 2014 Presentation and Gradiant Interview: Helena Fernandez Lopez. Friday February 21 2014 at 16:00.

Gradiant could therefore offer a personalized service to ASUK: the company could act as a consultant and advisor but could also create a personalized prototype solution tailored to ASUK's demands. This could in turn be manufactured and reproduced by one of their partners. Moreover, Gradiant has the technology know-how to provide a variety of solutions: providing services for patients with dual sensory loss, contact-less monitors of breathing and heart rate...Gradiant furthermore provides a sensible competitive advantage in that its 2020 strategy is to increase its participation in Horizon projects with their knowledge of the rules and fundraising tools but also their regular presence in most of the relevant European Technology Platforms. This will help not only ASUK in its implementation of a virtual clinic but would also be easily transferable to other rare disease charities or MDCs in Europe and the world.

The apparent limitations of Gradiant are firstly and most importantly that it does not outwardly offer a pre-made platform where the physicians, specialists and patients can interact in a virtual method, such as the healthcare islands of IBM. Gradiant is a consultant, advisor and prototype researcher, but does not manufacture or produce any products. This is a drawback, as it takes away the entertaining and reassuring aspects of having an avatar, which can represent you (helps personalize the virtual aspect), which you can control, and via whom you can interact with others. Gradiant can only provide technical knowledge but cannot offer a physical platform on which to implement it – that would result in extra costing and work for ASUK. This is a crucial aspect that differentiated IBM from Gradiant's offer. Moreover, Gradiant is currently working on multiple projects with numerous partners, which might hinder their availability and responsiveness, not only to transfer their eHealth knowledge but also to adapt their pricing and costs to those demanded by ASUK. Lastly, there is no mention of monitors or applications that patients can use to draw tests themselves and therefore reduce their over costs, time and travelling expenses to consult with their physicians.

Cisco Connected Care³

Cisco Systems is an American firm that designs, manufactures and sells networking equipment. Cisco's focus on three market segments relating to enterprise and service providers, small business and the home. In 2009, Cisco and UnitedHealth group jointly showcased their "connected care" mobile clinic that links patients to healthcare services. This first national telemedicine network has the goal of delivering high-quality healthcare services in different locations (rural, retail and workplace) where it is difficult to organize physical consultations with physicians or specialists. Cisco Connected Health therefore supports mobility and security with advisory services, security services, architecture assessment services and optimization all meeting cost targets. The virtual clinic contains care-at-a-distance clinical and workflow solutions for physicians and patients that are examined.

Cisco's virtual healthcare offer is outlined below.

³ http://www.cisco.com/web/strategy/docs/healthcare/11CS3289-ConnectedHealth_AAG_R2.pdf
and https://blogs.cisco.com/news/cisco_healthpresence_telemedicine_mobile_clinic_on_a_roll/

Offer	Cisco Connected Care
Platform	<ul style="list-style-type: none"> Care-at-a-Distance virtual platform
Services	<ul style="list-style-type: none"> Virtual consultations Nurse integration (communication with specialized phone) Network access control Extensive network security
Devices/ Monitors	<ul style="list-style-type: none"> Lightweight and mobile integrated video solution Specialized phone: real-time location (information and video), bar-code scanning, custom applications
Pricing	<ul style="list-style-type: none"> No pricing information communicated

Figure 24 eHealth offers: Cisco

Cisco is therefore beneficial for ASUK’s goal of implementing a multidisciplinary virtual clinic. It provides, unlike Gradiant, the physical technological platform on which the patients and physicians can interact. This platform contains all the necessary technical and network requirements to implement the virtual clinic through video and mobile devices, an installed and broad network as well as the security necessary to protect all the members of the virtual clinic. These features would allow a significant time and cost reduction for patients of ASUK, physicians and specialists as it would reduce travel and transport expenses, consultation times, hidden patient medical records that are not always available to all the physicians and lastly the patient benefit of feeling supported and followed.

Although Cisco contains the necessary requirements to implement a virtual clinic as it offers not only the product but also the network and security, it does have some apparent limitations. The first limitation is that Cisco is an American company operating in San Francisco, and although one of their virtual clinic pilots took place in Scotland and France, the firm is still based in America and not Europe. Moreover, although Cisco does provide products and devices, it does not seem to have integrated medical applications in their offered virtual clinic nor supporting devices for patients suffering from dual sensory loss like those in ASUK. Lastly, unlike IBM, Cisco does not offer an entertaining and original platform on which the physicians and patients can interact, losing the modern, fun and personalized aspect of the online virtual clinic.

Vodafone M2M⁴

Vodafone is a British multinational telecommunications headquartered in London and is the world’s third largest mobile telecommunications company. Although Vodafone’s business focuses mainly on telecommunications, it has a section dedicated to eHealth services. Vodafone created a “machine to machine (M2M)” program dedicated on improving, finding solutions and implementing eHealth platforms to its clients. It provides its solutions in three core areas: condition management, hospital to home and assisted living by not only addressing costing challenges but also increasing the quality of life of patients and physicians.

The offer for Vodafone’s M2M service is as follows:

⁴ <http://m2m.vodafone.com/discover-m2m/industries/health/>

Offer	Vodafone M2M
Platform	<ul style="list-style-type: none"> Machine-to-Machine virtual platform
Services	<ul style="list-style-type: none"> Unobtrusive remote monitoring and control of patient conditions in home Real-Time data access and exchange (diary) Digital exchange (virtual consultations)
Devices/ Monitors	<ul style="list-style-type: none"> Home safety monitors Blood Pressure Blood Sugar Vital sign monitor Data Interface in mirror cabinet (bathroom) Out-of-home diary
Pricing	<ul style="list-style-type: none"> No pricing information communicated

Figure 25 eHealth offers: Vodafone M2M

Vodafone's M2M service has numerous benefits for ASUK and also for other rare-disease organizations that require eHealth. It provides a three-step solution in condition management, hospital to home and assisted living. Vodafone delivers therefore an end-to-end solution that includes a fully managed connectivity, a global M2M platform to monitor and manage all the services, a logistics team that can assist in forecasting and lastly provides comprehensive support. Vodafone provides this service by first consulting with the clients to understand the requirement and devices necessary; then Vodafone provides a solution design and development, implements the platform and lastly monitors the functioning of the eHealth platform with its operational services. This is highly beneficial to Alstrom UK, as unlike Gradiant, Vodafone is responsible for everything in the implementation of the virtual clinic through a personalized consultation. Moreover, Vodafone has already tested monitoring devices for symptoms such as Diabetes, which is one of the symptoms of Alstrom UK patients. Lastly, Vodafone is based in the United Kingdom providing on-spot service in English.

Although Vodafone has a promising platform, there are some limitations in the offer. Firstly, there is not mention on how they could create sensors and devices for patients suffering from dual sensory losses (requiring advanced technical knowledge). Secondly, although Vodafone does provide a fully integrated platform, there is no mention on how it works across borders. Vodafone is a British telecommunications system that does not mention operating in other countries in Europe. If the platform is only maximized when utilized in the UK, it will inhibit cross-border specialists to monitor patients and participate in the process. This is an important limitation as most specialists of the Alstrom disease and other rare-diseases are scattered around Europe and on other continents. The platform needs to, therefore, be implemented internationally in order to reunite the best medical professors to monitor and advice on the health of the patients.

Orange⁵

Orange is a French telecommunications service provider that is globally implemented serving 33 countries with direct mobile presence and providing services to 231mn customers worldwide (Orange presentation, 2012). Orange has strongly developed, in line with their global innovation strategy, to create an integrated international eHealth service that provides applications and

⁵ Orange's Offer Presentation to ASUK

monitoring devices to tailor the needs of patients and physicians in countries ranging from the UK to Kenya.

Orange Healthcare offer is outlined below:

Offer	Orange Healthcare
Platform	<ul style="list-style-type: none"> • Integrated virtual platform
Services	<ul style="list-style-type: none"> • Connectivity and Data collection • Community portal • Collaborative workspace • Social Networking Facilities • Document and Data sharing
Devices/ Monitors	<ul style="list-style-type: none"> • Instant Video • Multichannel messaging and alarms • Orange Intelligent App • Monitoring Devices
	<ul style="list-style-type: none"> • Virtual Clinic Building Costs: 985 000 euros • Operation costs: <ul style="list-style-type: none"> ○ 15 045- 20 090 euros (monthly) ○ 450-1200 euros one-off user costs

Figure 26 eHealth offers: Orange

Orange healthcare’s solution provides numerous potential benefits for ASUK. Firstly, Orange provides a fully integrated platform that connects cross-border patients with physicians, physicians amongst themselves and monitors patient symptom escalations virtually. Orange furthermore builds on this platform using: feature highlights including collaborative workspace, functional architecture with an integrated network and technical assets using applications and devices to keep the participants connected. The most important benefit of Orange Healthcare is their fast response and readiness to deliver a researched integrated service plan specific to Alstrom UK. Orange has provide a tailored solution based on the needs and requirements of Alstrom patients, based on the charities time frame and costing budgets in order to produce the best fitted solution. This rapid service is something that can be applicable to all other rare-disease charities or players that require a detailed project plan to assess the costs and benefits of implementing a virtual healthcare program for their patients. Lastly, Orange is a global player that can truly promise to connect patients and physicians living beyond Europe in order to provide the best knowledge hub and integrated care platform.

The most apparent limitation rom Orange’s offer is the lack of mention on how they could create sensors and devices for patients suffering from dual sensory losses (requiring advanced technical knowledge). This however, can be built using various specialists in technology, app creation and the medical body. Moreover, although Orange has provided ASUK with pricing information, it is difficult to assess its competitiveness due to a lacking benchmarking model.

Case Study – Benefits of eHealth with Vodafone M2M Trial ⁶

Vodafone UK conducted a trial to test the efficiency and benefits of the remote monitoring of Diabetes type 1 patients with their device. This trial is relevant to ASUK and other charities of rare diseases, where diabetes can be one of the symptoms of their patients.

Purpose: Vodafone and a corporate partner sponsored this nine-month UK-based trial to discover how continuous monitoring of patients with mobile communications could help patient's manage their health and symptoms. Patients were therefore provided with a mobile application allowing them to securely log their test results (sent directly to the medical staff).

Sample: 93% of patients aged between 18-30 years with HbA1c levels of 8-11% took part in the study. These patients were given a "One Touch Ultra glucose monitor" and the mobile phone app allowing them to record their insulin level, food intake and activity. The data was then sent through a remote server to a data-processing facility. The sample was separated into a control group and intervention group.

Intervention Group: The intervention group received their data in the form of a time series of the previous 24 hours showing a color-coded histogram displaying results within the target range of the previous two weeks. The readings were monitored and assessed by specialist diabetes nurses at least fortnightly. Nurses also intervened and called patients to identify concerns, problems and collaborate to find solutions through the shared analyses. Treatment plans encouraged patients to adhere to multiple insulin injections when appropriate.

Control Group: The control group logged their test results in the diary but was not provided any medical support. They were sent feedback on their input via a times series graph showing the previous 24 hours and could access their own diary through the Internet. They had not medical support or advice on their treatment plan.

Results: The intervention group benefited from improvement in blood sugar levels and a reduction in HbA1c. The patients in the intervention group tended to adhere more strictly to the testing regime with the continuous support than the control group.

The patients in the intervention group also communicated their perceived benefits of feeling safe and supported; having closer communication with nurses, better treatment compliance and health outcomes, and lastly an easy and discreet management of their condition avoiding escalations of the symptoms.

The healthcare providers' benefits in this trial included maintaining a higher quality of life for their patients, a resource-efficient way to manage large population of patients, and fewer secondary care admissions caused by escalating health conditions.

Post-trial: Following the benefits of the study, Vodafone provided a commercial solution for diabetes condition management as part of their eHealth Solutions portfolio.

⁶ Vodafone eHealth Solutions Presentation from Remote Care Services (Condition Management): Condition management Remote Monitoring of Diabetes Type 1 patients.

Conclusion

In order to assess the offers described above, we have created a table depicting the five providers and their match to the important offer criteria for ASUK's eHealth system:

Offer Criteria	IBM	Gradiant	Cisco	Vodafone	Orange
Pricing	✓				✓
Services/Devices	✓		✓	✓	✓
Adaptability to Alstrom Syndrome		✓			✓
Availability				✓	✓
Cross-Border Potential		✓			✓

Figure 27 Offer criteria table

In the table, the most relevant choices for Alstrom UK would be firstly Orange and then Vodafone. The first reason is that IBM and Cisco are both operators based in the United States, making the implementation and monitoring of the virtual clinic more difficult with distance. Furthermore, these two operators have been difficult to reach for pricing. Moreover, Gradiant does not offer any product or platform but instead prototyping and consulting services, which are not adequate for ASUK's quest of implementing a virtual clinic. Orange and Vodafone, on the other hand, are both based in Europe and offer a complete eHealth service from consultation and prototyping to implementation and monitoring. Orange however, remains the leading competitive offer due to its tailoring to ASUK, its complete project pricing and duration, and its competitive devices suited for the needs of Alstrom patients.

The limitation of this analysis regards the pricing, the eHealth personalization of applications, software or devices, and the subjective criteria in electing the best-suited offer for ASUK. Most of the providers were incapable of giving a set pricing for their virtual clinic claiming that it is case-based including hourly fees, which depends on the depth and length of the project. Therefore, it is difficult to assess the offers based on economics and budget. Moreover, many of the virtual clinics proposed offer a variety of applications and monitors that are not adapted to patients suffering from dual sensory loss. This is a limitation as some providers do not have the ability to overcome this symptom or they can modify the devices at a greater cost. The criteria used to elect the best option are a subjective account of what is reported to be most important to ASUK, to patients, to physicians and lastly to the overall budget. These are freely interpreted but are still used as a benchmark in deciding which option seems to be the best suited and most complete for ASUK and for other potential rare-disease charities that would be interested in implementing the virtual clinic concept.

Alstrom as a paradigm rare disease

Investigation into the needs of other rare disease charities

Background & Methodology

In ASUK's case, MDCs are a successful step to consolidating treatment and knowledge with the aim of treating the disease's various symptoms. Aided by a combination of information and communication technology, the new model of virtual multi-disciplinary clinics is another leap towards effectiveness and efficiency.

Apart from AS, there are numerous other rare diseases and multi-symptoms disorders that require a multi-disciplinary approach to improve research, treatment and management. According to Rare Disease UK (RDUK)¹², there are over 6,000 recognized rare diseases, affecting approximately 3.5 million people in the UK. Registered members of RDUK exceed 1,400 and include academics, clinicians, industry, individual members and patient organisations.

There is great potential to benefit other rare disease organisations by applying ASUK's multi-disciplinary model. However, given the complexity of disease-specific symptoms; causes, treatments, and patient demographic characteristics make it difficult to simply promote ASUK's model without customizing the offer based on specific patient and physician needs. We have therefore investigated other rare disease charities regarding their current practice, their attitude and specific needs in relation to MDCs and the use of eHealth technologies to determine how ASUK's proposed business model might be replicable.

First, we selected 5 charities from over 500 rare disease charities and patient organizations based on the following criteria:

- The rare disease had to be a syndrome with multiple symptoms that shared some similarities with AS, and its treatment therefore calling for multi-disciplinary practices.
- The charities chosen are formal, experienced and either regional or national, resulting in a need for eHealth technologies to facilitate remote communication, monitoring and diagnosis.

Charity Name	Charity Profile	Disease Symptoms
Ataxia-Telangiectasia society (A-T society)¹³	Founded in 2004 Employees: 3 FTEs Patients: 167+ Medical resources: connected to 25 professionals & 2 hospitals Focus on UK service	Cardiomyopathy Neutropenia Fatigue and general muscle weakness Growth/feeding issues
The Barth Syndrome Trust¹⁴	Employees: 4 part-time volunteers Patient families: 20 Medical resources: work closely with 8 professionals & 2 hospitals Have international coverage	Difficulties in controlling eye-movements difficulties in pronouncing words clearly difficulties in swallowing which in turn can lead to loss of weight and to drooling Serious lung disease Postural problems & on occasion scoliosis of the spine

¹² Rare diseases UK - <http://www.raredisease.org.uk/members.htm>

¹³ Ataxia-Telangiectasia society (A-T society) - <http://www.atsociety.org.uk/home>

¹⁴ The Barth Syndrome Trust - <http://www.barthsyndrome.org.uk/>

<p>Vasculitis UK¹⁵</p>	<p>Founded since 1992 Employees: 5-6 FTEs Patients: 1100 in database Medical resources: 2 medical advisors, work closely with 8-10 hospitals Focus on UK service</p>	<p>Respiratory system : breathlessness, wheeze, dry cough or coughing up blood ENT (Ear, Nose and Throat) : hearing problems, nasal crusting, nose bleeds, sinus pain or hoarse voice Skin - rashes, ulcers, and necrosis Eyes - red eyes, painful, dry or gritty eyes, visual loss or other changes in vision Joints - arthralgia, and joint swelling Nervous system - loss of sensation, weakness, unusual pain in the hands and feet and rarely paralysis or stroke Gastrointestinal system - diarrhoea, bleeding & abdominal pain Kidneys/Renal - problems with minute amounts of blood or protein in the urine.</p>
<p>Meningioma UK¹⁶</p>	<p>Founded in 1997 Employees: 30 Patient: 2000~ Medical resources: connected to approx. 100 professionals & 100 hospitals UK national wide coverage</p>	<p>Bone disease Recurring infection due to treatments interfere with the immune system Anaemia: a reduction in the number of red blood cells and can cause fatigue, weakness or breathlessness Kidney damage caused by the myeloma itself or as a side-effect of treatment Hypocalcaemia: the level of calcium in the blood is too high and can cause thirst, nausea, vomiting, confusion and/or constipation Peripheral neuropathy: damage to the nerves that make up the peripheral nervous system.</p>
<p>Lymphangiomatosis & Gorham's Disease Alliance - Europe¹⁷</p>	<p>Founded in 2010 Employees: 5-6 FTEs Patients: 1100 in database Medical resources: 2 medical advisors, work closely with 8-10 hospitals EU-wide coverage</p>	<p>Pericardial effusions (fluid around the heart), Pleural effusion (fluid around the lung cavity), Ascites (fluid in the abdominal cavity), bone fractures, skin lesions, fever and internal bleeding. Shortness of breath, cough, difficulty breathing in (inhaling), severe pain in the abdominal cavity and pelvic cavity, lymphedema (swelling).</p>

Figure 28 Charities overview

We reached out to the 5 charities and four¹⁸ of them agreed to participate in our research via face-to-face or Skype interviews combined with a questionnaire on the “Evaluation of Multi-Disciplinary Clinics & eHealth system on Rear Disease Charities”. The interviews and questionnaires were designed with the aim to:

- Get general information on those charities
- Explore the levels of access to and usage of MDCs and eHealth technology in these charities

¹⁵ Vasculitis UK - <http://www.vasculitis.org.uk/>

¹⁶ Meningioma UK - <http://www.myeloma.org.uk/>

¹⁷ Lymphangiomatosis & Gorham's Disease Alliance – Europe <http://lqdalliance-europe.org/index.html>

¹⁸ Lymphangiomatosis & Gorham's Disease Alliance staff are on their winter vacation and may respond later on

- Identify the potential benefits of as well as the barriers to setting up MDCs and applying eHealth technology to aid the diagnosis, treatment and management of the specific rare disease
- Ask for recommended ways to maximize the benefits and to overcome barriers linked to MDCs
- Identify their attitude and willingness to adopt and invest in ASUK-like MDCs and eHealth technology systems

Key Findings

Multi-disciplinary clinics Part

High recognition of multi-disciplinary clinics but low levels of Implementation

All charities agreed that a MDC service is important for both the diagnosis and the treatment of their disease. Although there was one outlier regarding the gap between the very patient-faced clinical treatment and the lab-based medical research, 75% of the interviewees believed that such an approach is helpful for disease research as a result of better data collection and patient understanding.

Despite the high MDC recognition rate, only 2 charities (A-T Society & Barth Syndrome Trust¹⁹) have implemented these clinics in the past. Unfortunately, these two clinics were only in action short-term as a result of low funding and key staff departure. Although the other charities have not implemented MDCs, they still encourage this approach and willingly provide information to patients and healthcare professionals.

The one-stop clinic service is more timesaving, holistic and accurate

“Reduce clinic individual hospital visits/appointments” is probably the most direct benefit for patients (100% vote) and for physicians/specialists with time efficiency (75% vote). Another important benefit brought by the combination of multi-disciplinary medical team is to *“increase accuracy and reduce misdiagnoses”*. This is a result of a more holistic view of a patient’s profile - especially when some symptoms are similar to other diseases. Lastly, having an integrated platform uniting all relevant specialists leads to a better coordination and knowledge sharing resulting in a more holistic care.

Coordination and finance are the biggest challenges

Although the multi-disciplinary clinic approach is largely supported by charities, it is *“time and labour consuming in coordination and organization”* given that most charities are small in size. What is more, regularly operating the MDCs is also a big and continuous financial challenge. Even when funded by the NHS, the charities find it difficult to expand their client base. Long-distance travelling and accommodation are also indicated as large barriers to setting up and controlling MDC.

More fundamentally, there is still a gap between demand and supply. The real demand for MDCs is underestimated due to the lack of patient knowledge regarding the existence of such platforms. On the other hand, only a few clinics provide and supply a high quality multi-disciplinary clinic. Most local clinics do not have a comprehensive multi-disciplinary team and lack cooperation with local hospitals.

¹⁹ Barth Syndrome Trust’s case is quite similar to ASUK, in which the charity was the main initiator at first and clinics were held with voluntary help from medical personnel involved. But since April 2010, funding was obtained from NHS for a specialized service. A-T Society started such practice very early in 1994 but stopped for quite a few years when the key staff left, then re-opened in 2002 & 2008

Weighing the benefits and limitations, two out of four charities claimed that they would not be highly involved in implementing a multi-disciplinary clinic.

eHealth Technology Part

Technology can help in many ways

According to the interviewees, eHealth technology can help patients, professionals and hospitals in several aspects. In the top 5 listed below, *information, communication, cost saving, data and safety* are the predominant reasons:

- Access to online information, evidence and guidelines
- Better communication/interaction to support patients
- Save time & cost for patients (e.g. travel expense)
- Support safer patient care (e.g. emergencies that could have been avoided with continuous monitoring)
- Improve nursing or treatment decision making (with more comprehensive data and better reporting features)
- More data available to help research

Uncertainty in attitude towards eHealth Technology

Although all of the charity leaders said new technology in eHealth is beneficial, they remained sceptical of how fast the relevant technological improvements will appear. Out of those interviewed, two interviewees thought that the application of eHealth technology was very important, while the two others were unsure (“maybe” and “in between”).

Limited access and usage of eHealth technology and lack of customization

75% of interviewees admitted that the “*level of access patients have to eHealth system /ICT equipment is far from adequate*”, whilst the other 25% believed it to be moderate. The top three eHealth solutions in importance are “Online information & social networking” (100% vote); “Communication tools to increase patients/physician interaction or remote diagnosis”(75% vote); “Electronic medical record (EMR) system or central online database”(50% vote). Almost **no** (wearable/smart) devices for measuring and monitoring and specific technology used for new diagnostics were mentioned. One interviewee indicated that a few patients used mobile/portable commercial apps (downloadable from the App store) for personal health management but these lack medical precision and customization to specific rare disease symptoms.

Regarding the question “What can be the eHealth solution/consultation that you or your community want most?” *Information & knowledge access, special test device, co-cooperative tools and telemedicine* were the top votes. Requirements for this include:

- Increased access for patients to their medical records online.
- A Knowledge sharing or e-learning platform, especially increased access by generalists dealing with patients to specialists/experts e.g. case conferences, education material
- Electronic results of diagnostic tests
- More online telemedicine without the constraints that NHS places on hospital staff who cannot access VOIP from hospital due to security issues.
- More Skype access between medical staff and families.
- Better devices needed to monitor health at home (e.g. heart rate monitors, blood glucose monitors, special blood test device for home use.)

Lack of financial and technical support is a major barrier

“Lack of technical support to develop or setup eHealth” and “Lack of financial support to invest in eHealth” were indicated as the top two barriers (100% vote) that hinder the promotion and the adoption of eHealth technology for rare diseases. Current technological solutions cannot meet all the specific demand from the patient side. Given the wide range of symptoms in those syndromes, the cost of developing an integrated technological solution can be very high. Moreover, the development of such technology may not be scalable and many firms lack the commercial incentives to research and develop these.

Other challenges such as the “lack of a systematic/integrated tech solutions & consultation”, the “lack of collaboration among different stakeholders”, the “lack of efficient patient education” as well as “concerns over data and privacy” should not be overlooked.

In the long term, 75% of the charities project that the benefits of using eHealth will outweigh the costs.

Communicator and connector are major roles that charities are going to play

In response to the question “In what aspect(s), do you think rare disease charities or patient organization can help promote the eHealth concept and technologies?”, all interviewees asserted that their charities could help most in “educating and facilitating the patients to adopt the eHealth concept and solutions”.

Moreover, 75% of charities positioned themselves as the effective connector and communicator between patients and physicians. Similarly, 75% of these charities would like to cooperate with technological providers by inputting their knowledge to help the development or customization of eHealth system. Two charities further expressed their willingness to help collect and process patient data in the elementary level.

As for charities’ investment in a systematic eHealth technological solution, only one charity said they would try to invest more within their budget. Others were unsure; most of them believed it should be the government or big technological companies’ responsibility to finance or sponsor in order to have a more effective and wider impact.

Conclusion

From the investigation’s findings, we can draw the following conclusions:

1. **The multi-disciplinary clinic service, highly encouraged by charities, has been introduced to other multi-symptom rare diseases as a one-stop, holistic and effective care service for patients.** Despite the proven benefits to patients and physicians, the holding of multi-disciplinary clinics is constrained by time, place, money and people. Building a comprehensive team of multi-disciplinary specialists within one hospital is not going to impact the whole client base, reducing advantages and benefits.
2. **eHealth is a promising trend** that will largely improve both the efficiency and accuracy of treatment as well as patients’ self-management and satisfaction. However, its development is still at an early stage; conceptual and not systematic in most charities’ opinion. Generally, most patients, charities and even some medical professionals have not yet adopted and show little usage of eHealth technologies. The reasons are as follows: limited knowledge

about technology (many of them are not technologically-savvy), and the lack of right technologically tailored implementation for rare diseases at an affordable price.

3. **Compared to ASUK, many similarities can be found among other rare disease charities.** Charities all expressed a willingness to support and promote multi-disciplinary clinic service and eHealth with recognition of its benefits. But in reality, investing in these new solutions requires large investments, expertise and cross-border collaboration. Charities' participation and impact can be limited given their size, knowledge and resources. Besides, some concerns, such as those surrounding privacy and technological abuse, still remain important. Moreover, different rare diseases vary from one another and from patient to patient, adding difficulties for other disease charities to replicate ASUK's model. Thus not all of the charities will fully embrace those new solutions until the relevant technology becomes more mature.

Based on the above conclusions, we have come up with some recommendations:

1. **The new model of virtual multi-disciplinary clinics that combine the multi-disciplinary clinic approach with the eHealth technology can be useful in the treatment and management of rare diseases.** eHealth technologies, such as remote monitoring, EMR and co-working platform, may be a good complement to multi-disciplinary clinics given its real-time, data-driven and remote features, and can increase patients' access, improve diagnosis as well as intensify collaboration among stakeholders.
2. **National and international public service providers, such as the NHS and EU, should be in charge** of the investment, promotion and regulation in the development of MDCs and eHealth technology. More specifically, *a central multi-disciplinary specialist team* should be setup, providing guidance, training and support to local hospitals and clinicians. At the same time, *more incentives should be provided to IT giants* to motivate them develop the necessary technology for rare diseases.
3. **Charities can act as change agents**, which are actively involved in supporting and promoting multi-disciplinary clinics and eHealth technology even beyond the roles of evangelist and connectors. They can come up with clear vision, advocate changes actively, leverage their relationship in the community, and share knowledge by leading by example (e.g. take advanced tools at first). With such a position, charities can be an indispensable part for the evolution of rare disease treatment.
4. **ASUK could step forward to initiate the changes.** Given the complexity and difference among rare disease charities, we would *provide consultancy over a systematic virtual multi-disciplinary clinic to other charities as a better way to leverage insights and demonstrate great leadership*. The consultancy can cover the best practice sharing, staff training and recommendation of technological solutions. A toolbox could be built accordingly.

Appendix

Appendix

Exhibit 1: Approach

Two approaches can be used to calculate the economic costs associated with Alstrom Syndrome; a prevalence approach, which is the "stream of medical costs attributable to all patients alive with the disease during a specified time period," and an incidence approach, used more commonly for acute disease, and which is "the present discounted expected sum of current and future costs accruing to all incident cases in a time period" (Meiners & Hodgson, 1982). This study uses a prevalence-based approach.

Costs are primarily categorized into direct costs and indirect costs. Direct costs are the value of all resources used in the medical treatment of the Syndrome or its effects. These are determined on a pre-multidisciplinary clinic basis as well as on a post-multidisciplinary basis, to evaluate the effects of the transition.

Indirect costs are those not attributable to the medical treatment or management of the syndrome. Instead they are the socio-economic implications of Alstrom Syndrome. For this study we analyze the financial, economic and emotional impacts of living with and caring for somebody with Alstrom Syndrome. Again, this is done on a pre-multidisciplinary clinic basis as well as on a post-multidisciplinary clinic basis. Economic costs of premature mortality as well as gains or losses attributable to morbidity are evaluated and used to draw inferences regarding the consequences of multidisciplinary clinics.

Due to low prevalence of Alstrom Syndrome, little data exists to provide reliable estimates of health care utilization, expenditures, mortality costs and morbidity impacts. National surveys are the usual source of data for cost of illness studies. However, because these surveys generally sample a very small proportion of the population, they do not contain enough persons with rare diseases to provide reliable estimates. To compensate for this, we used other sources of data to make our analysis. These included costing models drawing off national hospitalization rates, patient/care giver surveys as well as extensive patient/care giver interviews and case studies.

Exhibit 2: Methods and Data Sources

Cost Models

The initial idea for building the costing model was to compare the costs prior to the MDC, the actual MDC costs and the Virtual MDC costs.

However, after discussing the approach with the physicians responsible for the MDCs in Birmingham and with a Finance Manager from NHS, it became clear that the approach had to be changed for several reasons.

One reason was that the information we were looking for is not available in that format. Our approach assumed that the NHS had a cost figure for every treatment that is received by patient. So for example each blood test would cost the NHS a certain amount of money. However, the way the costing takes place at NHS that hourly rates for specialists are calculated and that then in a separated step the costs for materials are calculated. Therefore we adjusted our approach, starting from the hourly rate of the specialists involved and estimating the material necessary.

Another reason for the adjustment was that the physicians made clear that the eHealth application at discussion would be used on top of the MDCs, i.e. the costs for the MDC would still be same.

However, one could for example assume that by using eHealth applications the number of visits could for example be reduced. Therefore, we are setting the costs for the MDC in relation the costs (prices) provided by the eHealth application providers, in order to analyze a potential cost benefit of the eHealth application and in order to check the reasonableness of introducing the eHealth applications.

Interviews

Fundamental premises

Now turning to the qualitative research methods employed in our analysis, our approach was twofold; first, we conducted a series of interviews with several Alström patients, and used the initial information gained and insights provided to design a tailor made survey to increase our sample size beyond the limitation of interviewees (see below).

Our reasoning for conducting the interviews was based on several calculations, not least our desire to get a better understanding of our patients' life with AS, their treatment needs and previous experiences with treatment methods – both in ASUK's multi-disciplinary clinics and outside them. Although "Qualitative methods occupy an ambivalent position in social care research" (Moriarty, 2011: 31), this approach is confirmed by Kvale and Steinar (1996). The latter claim that the objective of an interview lies in "obtaining qualitative descriptions of the life world of the subject with respect to interpretation of their meaning".²⁰

What is more, we did not wish to limit our insight into the condition purely to medical articles and statistics prior to designing our survey, and in this context the interviews provided us with a good understanding of real life issues with Alström treatment methods. These could, in a further step, be validated in the survey.

The main advantage of interviews according to Moriarty is the vast amount of data that can be collected in a limited amount of time – a point that we can only confirm. However, set up time and the efforts after the interview to write up the data collected and analyse the results and implications should not be underestimated.

Interview structure

Interviews in qualitative research are one of the most common data collection methods and may follow various structures. Two of the most of common of types of interviews are:²¹

1. Semi-structured interviews: These are built around a number of open and semi-open questions that are posed by the interviewer. The interviewer thereby chooses the topics he or she thinks relevant, and in the process can cover a number of issues.
2. In depth interviews: As opposed to the more broad semi-structured interview setup, in-depth interviews only cover a small number of topics (perhaps even only a single issue). However, the level of detail is far greater. What is more, in-depth interviews allow the researcher to validate theories he or she may have and make it possible to benefit from the full level of expertise the interviewee may have.

As the main aim of our initial interviews was to further our understanding of the patients' experience with AS and in the MD clinics, we chose a semi-structured interview set-up. Prior to the interviews, we drafted a broad list of questions that we were able to draw on in our talks with the

²⁰ Kvale, Steinar (1996): Interviews An Introduction to Qualitative Research Interviewing, Sage Publications.

²¹ Moriarty, J (2011): School of Social Cares Research: Qualitative Methods Overview, National Institute for Health Research, London School of Economics. London, UK.

patients/carers, but it is safe to say that the direction of the interview varied from case to case, depending on the interviewees' views, feedback and conditions. We were thus able to gain a broad understanding of individual circumstances. What is more, we did not have a hypothesis that needed verifying, making the in-depth structure inappropriate.

Location and interview set-up

We recognise that a face-to-face setup would have ideally been the best option, as these have several "advantages in terms of creating rapport and also allows researchers to observe participants' non-verbal communication, such as their use of gestures" (Moriarty, 2011: 9). However, due to time (the project has a tight deadline) and location (not all patients are based within London) constraints, we conducted telephone interviews.

Irvine, Drew and Sainsbury²² compared telephone and face-to-face interviews. One of the results they found was that telephone interviews last on average in their comparative study of telephone and face-to-face interview techniques, found that telephone interviews tend to be shorter in duration – a finding that we can confirm. The duration of our talks ranged from 35 to 60 minutes and was therefore less long on average than the length of the interviews that Moriarty cites, which lasted up to 90 minutes.²³

What is more, Irvine et al. (2010) underline the importance of giving continuous verbal feedback such as "yes" and "mm hm" when conducting telephone interviews in order to prove attentiveness to the interviewee; according to the author's research, these "verbal tokens" are more common in face to face interviews, and it is therefore important to actively give such tokens in a telephone interview – a piece of advice we did our best to follow. However, the researchers also claim that "participants in face-to-face interviews tended to speak for longer stretches at a time, before the researcher stepped in with another question or comment" (Irvine et al., 2010: 3). This was not, however, an experience we had, as all patients we spoke to were very forthcoming with information and generally seemed to appreciate the opportunity to convey their thoughts on the subjects discussed.

Surveys

Fundamental Premise

The survey used in this study played an important role in determining the socio economic implications of living with, caring for and treating Alstrom Syndrome.

The survey was set up in such a manner that we were able to use our findings in conjunction with other research done on the implications of rare diseases, across Europe as well as the United States. The purpose behind this approach was to review previous findings and dismiss or reiterate their results as well as to demonstrate that our conclusions are applicable across a number of rare diseases and syndromes. This tied in well with the rest of our report and became another case demonstrating the paradigm role Alstrom Syndrome can play.

In accordance with Fowler's (2009) definition of a survey, our survey met the following three criteria²⁴:

1. The purpose of the survey was to produce statistics
2. The main way of collecting the information was by asking people questions; their answers constituted the data to be analyzed

²² Irvine A, Drew P, Sainsbury R (2010): Mode effects in qualitative interviews: a comparison of semi-structured face-to-face and telephone interviews using conversation analysis. Research Works, 2010-03, University of York.

²³ Moriarty, J (2011): School of Social Cares Research: Qualitative Methods Overview, National Institute for Health Research, London School of Economics. London, UK.

²⁴ Fowler, F. J. (2009). *Applied Social Research Methods Series* (Vol. 1). (L. Bickman, Ed.) California, United States of America: SAGE Publications Inc.

3. Generally, information was collected about only a fraction of the population, that is, a sample, rather than from every member of the population.

The fundamental premise of our survey process was that by describing the sample of people who actually respond, we could describe the target population.

Survey Errors and Preventative Measures

Fowler (2009) identified two kinds of errors in relation to surveys. The first error of concern was that the results produced by the sample would vary and would not be a true depiction of the population²⁵. This variation error stems solely from the fact that data was collected from a sample rather than from every single member of the population. It is called sampling error.

For our study, the survey was distributed by way of the charity organization Alstrom Syndrome United Kingdom (ASUK) and their database of Alstrom Syndrome patients. Given the small number of known cases of Alstrom Syndrome in the UK and the fact that the ASUK act as equal partners in the provision of the syndrome's multidisciplinary clinics, our sample in effect targeted the entire UK population of Alstrom suffers. Ultimately, the UK survey results would be used to infer expected results of the European Union. Given the similarity in demographics across Europe and the fact the UK is a member of the European Union, we felt it provided a fair depiction of our target population. To ensure greater certainty, results from other similar studies were used to verify our data. As another precaution, a number of biographic and symptomatic variables were controlled when analyzing results (for example age, gender, symptoms suffered etc.)

The second type of potential error was in relation to bias²⁰. Bias means that in some systematic way people responding to the survey were different from the target population as a whole.

There are three steps in the process of collecting data that could introduce bias:

1. **Sample frame** – To avoid sample frame bias we set up our surveys so that the patient or their respective caregiver could complete it. For those who were unable to complete the survey due to its format, we offered the option of talking volunteers through the survey and completing them on their behalf. Assistants were strictly trained to ensure no bias was introduced during this process.
2. **Process of selecting** – Given the low prevalence of the syndrome, we attempted to reach out to all Alstrom Syndrome suffers across the UK. Our selection process involved no human discretion on the part of any of the researchers and instead attempted to reach as many patients as was possible.
3. **Failure to collect answers from everyone** – The survey was set up using an online digital form, which eliminated access problems relating to survey dissemination, completion and collection. To ensure everyone was able to complete the survey we provided support such as telephone assistance to work through the survey with patients who were unable to complete it due to varying sensory losses. We also ensured the survey was compatible with screen reader software such as JAWS, NVDA, ChromeVox, and VoiceOver. We also consulted with Proferros Claes Moller, MD PhD from Depå Audiology Universitet Hospital, Örebro in Sweden, who is an expert in the field of dual sensory losses.

When designing the specific questions of the survey, we took a number of measures to improve the their reliability (providing consistent measures in comparable situations) and validity (answers correspond to what they were intended to measure). The following measure were taken:

²⁵ Fowler, F. J. improving Survey Questions - Design and Evaluation. *Applied Social Research Methods Series* , 38, 7-24.

1. All respondents were asked identical sets of questions
2. Data validation parameters were implemented into the digital survey (in other words if a question required a numerical answer, only numeric inputs were accepted).
3. All questions needed to be answered in order to progress through the survey.
4. To avoid low return rates we partnered with ASUK to advocate the research we were conducting and to encourage those they support to contribute to our data collection. The survey itself was also simplified, shortened and made as unobtrusive as possible.
5. Surveys were kept completely anonymous and there was no ways of identifying the respondent. This was to ensure that all participants were willing to answer questions as accurately as possible with full confidentiality.
6. Careful attention was paid to how the survey was constructed:
 - a. Phrasing of questions were rigorously analyzed and refined (see discussion below)
 - b. Spacing of questions and section headings were designed to stimulate understanding and structure the survey to be more effective and intuitive.
 - c. Training of interviewers to avoid potential biases effecting responses. Interviews were provided a script to which they were expected to strictly adhere to.

When phasing questions, we ensured the design of each question overcame the following challenges²⁰:

1. Defining objectives and specifying the kind of answers needed to meet the objectives of the questions. This included putting in place specific formatting parameters.
2. Ensuring that all respondents had a shared, common understanding of the meaning of the question. Specifically, all respondents were ensured to have the same understanding of key terms of the question, and their understanding of those terms was the same as that intended by us as the research team.
3. Ensuring that respondents were asked questions to which they knew the answers, Barriers to knowing the answers took the following forms:
 - a. Never having the information needed to answer the question (Respondents were able to skip certain questions)
 - b. Having the information at some point but being unable to recall the information accurately (Questions used the phrases “Approximately” and provided bucket style answers options (e.g. between 1 and 5)).
 - c. Difficulty in accurately placing events in time
4. Asking questions that respondents are able to answer in the terms required by the question (It is possible to ask questions to which respondents literally know the answers but are unable to answer the way the investigator want because of a lack of fit between the desires of the investigator and the reality about which the respondent is reporting).
5. Asking questions that respondents were willing to answer accurately.

The survey was also phrased in such a manner that it avoided leading/biasing respondents in any way. Additional information was also provided to ensure every aspect of the survey was understandable and that this understanding was consistent across all respondents. Questions formats ranged from checkboxes, multiple choices questions, and range selection questions, numeric only as well as long answer paragraph style questions.

To view the final edition of our survey please [click here](#).

Testing and Distribution

Pre-distribution testing and reviews were also conducted. Mrs. Kay Parkinson of the ASUK was asked review the survey as well as circulate it amongst family support workers and lead physicians for their input. From this the survey was adjusted and recompiled and subsequently retested and reviewed. A final version was then created and made available for distribution.

The survey was distributed by way of the charity organization Alstrom Syndrome United Kingdom (ASUK) via their database of Alstrom Syndrome patients.

Samples Statistics

Our sample consisted of 14 respondents as was collected from the 9th March 2014 until the 26th March 2014. This represents an approximate 23% coverage of the entire UK Alstrom Syndrome population.

Limitations and Future Considerations

In the interest of timeliness, the survey took on a cross sectional design (these are usually employed when information about a population at one point in time is desired). However, the ideal design of the survey would have been longitudinal in nature, i.e. pre and post-multidisciplinary clinics surveys. This would allow an assessment of development and change over time. Overall in our situation this was not possible as the study only began in the post-multidisciplinary period. This creates an obvious difficulty in that it is not possible to gauge the validity of the retrospective answers given to the survey questions. However, as a future research proposal we recommend conducting longitudinal surveys for pre and post-eHealth multidisciplinary solutions – this will provide a rich platform of data to evaluate its implications²⁶.

Conduction Surveys and Interviews

We conducted both surveys as well as interviews for a number of reasons. Firstly, we used the structured questionnaires and semi-structured interviews as mixed method studies to generate confirmatory results (despite differences in methods of data collection, analysis, and interpretation²⁷). Secondly, while questionnaires help provide evidence of patterns amongst the larger population, qualitative interview data often gather more in-depth insights on participant attitudes, thoughts, and actions²⁸.

On-site visit of MDC in Birmingham

In order to get a better impression of what the facilities at the MDC look like and to get a better understanding of the process of the MDC on the two days, we visited the Birmingham Children's Hospital. Although the MDC did not take place on the day of our visit, we got a good impression of what the procedure will look like. This impression helped us for our analysis to be more feasible.

²⁶ Hackett, G. (1981, May). Survey Reserach Methods. *The Personnel And Guidance Journal* , 599-604.

²⁷ Brown, & Harris. (2010). Mixing interview & questionnaire methods. *Practical Assessment, Research and Evaluations* , 15 (1).

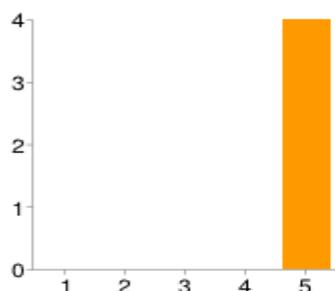
²⁸ Kendall, L. (2008). The conduct of qualitative interivew: Research questions, methodological issues, and researching online. In J. Coiro, M. Knobel, C.

Exhibit 3: Charity survey summary of Key Questions

Evaluation of Multi-Disciplinary Clinics & eHealth system on Rare Disease Charities.²⁹ The horizontal axis depicts the importance with which the factor was rated, and the vertical axis shows the number of responses obtained for that specific question.

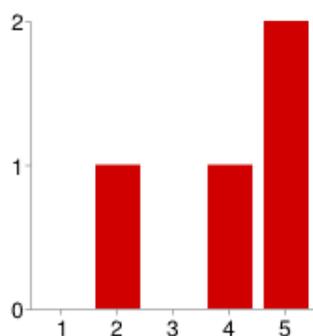
Part 2: Application of Multi-disciplinary Clinics

Is the multi-disciplinary approach important for disease diagnosis and treatment (On a scale of 1 to 5, with 5 being the most important)?



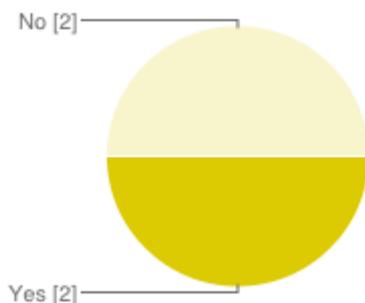
1	0	0%
2	0	0%
3	0	0%
4	0	0%
5	4	100%

Is multi-disciplinary approach important for disease research (On a scale of 1 to 5, with 5 being the most important)?



1	0	0%
2	1	25%
3	0	0%
4	1	25%
5	2	50%

Have your charity (as a main organizer) ever organised multi-disciplinary clinics or similar activities before?



Yes	2	50%
No	2	50%

In your opinion, what are the benefits of multi-disciplinary clinic services?

Increase accuracy and reduce misdiagnose	3	75%
Increase patients' access to healthcare resources	3	75%

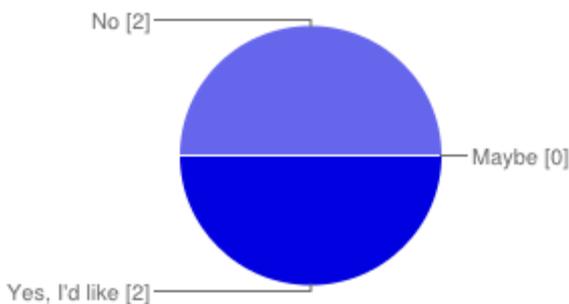
²⁹ The design of this survey has referred to RCN eHealth survey 2011 report by The Royal College of Nursing (2012) and A manifesto for a socio-technical approach to NHS and social care IT-enabled business change – to deliver effective high quality health and social care for all by Clegg C, Ellis B, Wyatt JC, Elliott B, Sinclair M and Wastell D (2010)

Reduce clinic individual hospital visits/appointments	4	100%
Time-efficient for the specialists and professionals	3	75%
Make patient feel more supported	2	50%
Better connection, interaction and cooperation in the community (even among the physicians)	2	50%
Good for collecting & managing patient data (for better research & monitoring)	3	75%
It's a bad idea	0	0%
Other	2	50%

What are the challenges or concerns for you to hold those multi-disciplinary clinics?

Patients and their families' accommodation during the clinics	1	25%
Coordination and organisation is time/labour consuming	2	50%
Lack of knowledge or experience to organize	0	0%
Difficult to finance such clinics regularly	2	50%
Long-distance travel needed for some patients & specialists	2	50%
Make little difference	0	0%
Other	2	50%

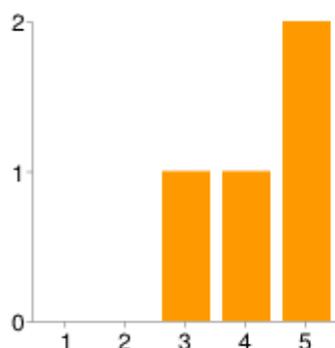
Are you willing to adopt multi-disciplinary clinic services in the future?



Yes, I'd like to	2	50%
No	2	50%
Maybe	0	0%

Part 3. Usage of eHealth system

Is the application of eHealth technology important? (On a scale of 1 to 5, with 5 being the most important, and the y-axis representing the number of respondents)

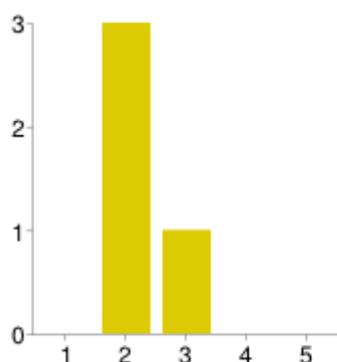


1	0	0%
2	0	0%
3	1	25%
4	1	25%
5	2	50%

What eHealth technology has been applied to the treatment and management?

Communication tools to increase patients/physician interaction or remote diagnosis (e.g. Skype/video-conference, mobile phone)	3	75%
Online information & social networking (e.g. online support website, patient forum, Facebook page, whatsapp groups etc.)	4	100%
(Wearable/smart) devices or sensors for measuring & monitoring (e.g. check weight, blood sugars etc.)	0	0%
Special mobile/portable apps for personal health management	1	25%
Electronic medical record (EMR) system or central online database	2	50%
New diagnostics technology	0	0%
Not applicable	0	0%
Other	0	0%

Do you consider that the level of access patients have to eHealth system /ICT equipment is adequate (On a scale of 1 to 5, with 5 being as adequate as possible, and the y-axis showing the number of respondents)



1	0	0%
2	3	75%
3	1	25%
4	0	0%
5	0	0%

What can be the eHealth solution/consultation that you or your community wants most? Please tell us more

- Increased access by patients to their medical records online or a facility that allows patients to enter and access information important to them electronically.
- Increased access by generalists dealing with patients to specialists/experts e.g. case conferences
- Educational materials for healthcare professionals that are available online or through apps e.g. e-learning
- Ability for healthcare professionals to share the results of diagnostic tests electronically, e.g. for case conferences, special blood test device for home use.
- More telemedicine via Internet without the constraints that NHS places on hospital staff who cannot access VOIP etc. from hospital due to security issues.
- More Skype access between medical staff and families. Better devices needed to monitor health at home (e.g. heart rate monitors, blood glucose monitors)

In which aspect(s) do you think eHealth technology can help for the patients, professionals and hospitals?

Access to online information, evidence and guidelines	3	75%
Better communication/interaction to make patients feel more supported	3	75%
Save time & cost for patients (e.g.travel expense)	3	75%

Support safer patient care (e.g. emergencies that could have been avoided with continuous monitoring)	3	75%
Support integrated/multi-disciplinary care	2	50%
Cost-saving for hospitals (e.g. investment/usage of medical equipment/resources)	2	50%
Improve how nursing care is recorded and reported	2	50%
Improve nursing or treatment decision making	3	75%
More data available to help research	3	75%
Other	0	0%

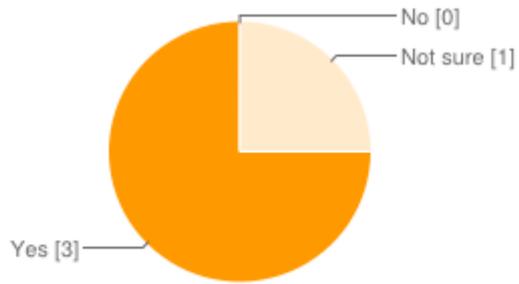
What are the challenges to promote the eHealth system using in rare diseases?

Lack of financial support to invest in eHealth system	4	100%
Lack of managerial support to adopt/promote eHealth system	2	50%
Lack of technical support to develop or setup the eHealth system, e.g. customization to the very specific diseases	4	100%
Lack of a systematic/integrated tech solutions & consultation	3	75%
Lack of collaboration among different stakeholders, e.g., data sharing	3	75%
Train the nursing staff/ families to use the system efficiently	2	50%
Educate the patients to use the system eHealth tech efficiently	3	75%
Concern over data and privacy	3	75%
Other	0	0%

In what aspect(s), do you think rare disease charities or patient organization can help in promoting the eHealth concept and technologies?

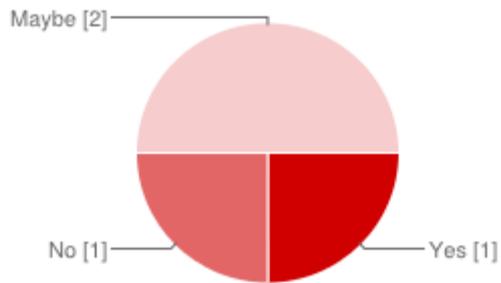
Provide knowledge to help the development of customized eHealth system	3	75%
Adopt and maintain an eHealth system that can be accessed by stakeholders/community	1	25%
Funding or subsidizing patients access to personalized eHealth solutions	1	25%
Educate and facilitate the patients to adopt the eHealth concept as well as the system	4	100%
Keep effective communication with the patients and physicians	3	75%
Help collect and process data in the elementary level	2	50%
Other	0	0%

Do you think the benefits of using eHealth will outweigh the cost in the long run?



Yes	3	75%
No	0	0%
Not sure	1	25%

Would you like to invest (more) in a systematic eHealth tech solution?



Yes	1	25%
No	1	25%
Maybe	2	50%