

# Development of a Comprehensive Care Plan for young Patients with Rare Diseases

## Introduction

In the past few years the view of rare diseases has changed markedly. It is now understood that people affected by rare diseases, though relatively small in number, not only require holistic care from an early age but that this care can actually be cost effective, minimising emergency treatments, as well as high social and educational burdens later on. Additionally, it is becoming recognised that investment in rare diseases advances knowledge in more mainstream conditions and increasingly has provided a focus for cutting edge research.

In the UK, the National Specialist Commissioning Group, (now AGNSS), has led the state NHS response to this need by funding clinics to focus resources on previously neglected rare conditions. Encouraged by Bruce Kent director of the Genetic Interest Group, the patient group Ålstrom Syndrome UK was a catalyst in this movement partnering with NHS centres of expertise at Torbay and BCH to set up the first essentially patient led specialist clinics in 2006/7. Since then, other similar clinics focusing on LMBBS, Wolfram's syndrome and PCD have been established following the model established by ASUK and their partners. By all measures this activity has been a resounding success, improving clinical outcomes, increasing patient wellbeing and sense of involvement, and providing the platform for outside funding for research into Alström Syndrome.

After 4 years of holding Alström syndrome clinics, the adult AS clinic at Torbay is in the process of being transferred to QEH Birmingham. This will bring it physically close to the Children's clinics at Birmingham Children's Hospital and expand the range of care options available to AS patients. It will also strengthen the ongoing move to make Birmingham a designated centre of excellence for the treatment of rare diseases. Coincidentally, we have also reached a point when children previously treated at a child clinic at BCH are ready to transfer to an adult clinic. Recently charities with common interests in rare diseases have begun to bind together to maximise opportunities for research and treatments. The Ciliopathy Alliance is one such grouping founded in 2010.

## Transition Processes

For rare and complex disease like AS, LMBBS etc, holistic medical treatment is essential for improving clinical outcomes. However to maximise patient life chances as well as minimising the overall 'whole life' cost to society we need to look beyond the purely medical care. As the child grows and matures the treatment and support needs change. In addition to the natural progression of the condition as patient age, which may require a changing mix of clinical expertise, passage from childhood through adolescence to adulthood brings social, sexual and educational challenges which are often magnified by the disease. While the introduction of the specialist clinics has brought

together clinical expertise across a wide spectrum to deliver holistic medical treatments, it has not attempted to address the wider social and educational needs which affect patient wellbeing and life chances nor the needs of the family whose support is so vital to the patient.

At ASUK we believe the congruence of factors mentioned above provides a unique opportunity to build on what has been achieved and raise our vision to another level where we consider how best to support children and families with a wide variety of challenging and complex conditions to improve not only health and wellbeing but maximise life chances overall. The objective is to allow these children to develop as adults maximising their individual potentials and, with the help of their clinicians can manage their medical conditions without being necessarily be defined by them. Furthermore, we believe that by mobilising other state functions and involving the charity sector in a shared goal, we can make a much greater impact without inevitably incurring enormous additional costs. We believe we should open our minds to solutions which have been shown to work in other countries such as the Ågrenska clinic model pioneered in Sweden and now embraced by Spain and Estonia<sup>12</sup>.

ASUK has shown leadership and vision in the past in pioneering the existing clinic model, securing and managing a research project and helping to found the Ciliopathy Alliance. We believe this equips us well to lead and coordinating this activity.

## Proposal

1. ASUK would take the lead in starting a wide discussion about the process of managing transition for all patients with rare, complex inherited diseases. (Note “transition” is shorthand for the overall process of supporting children (and their families) with medical, social and educational needs as they mature to adulthood.

2. ASUK partnering with BCH, QEH and TBH, will identify interested agencies such as;

Educational Establishments such as New College Worcester, Hereford College and possibly a University

Medical charities and patient support groups such LMBBS, Wolfram’s syndrome PCD, RNIB, Diabetes UK, GIG etc

Social Support charities such as National Centre for Independent Living

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1 [www.agrenska.se/en/Agrenska-model-for-center-in-Spain/](http://www.agrenska.se/en/Agrenska-model-for-center-in-Spain/)

2 [www.agrenska.ee/?setlang=15](http://www.agrenska.ee/?setlang=15)

Official bodies such as DHSS, AGNSS, Department of Education, Job Centre Plus (disability advisers)

Fund raising specialists

Etc,etc (ADD MORE TO LIST HERE)

3. ASUK will convene and chair meetings and discussion groups to

- Identify shortcomings in our present systems and evaluate opportunities for improvement, including evaluation of other approaches which have a proven track record of delivering better outcomes, (in particular the Agrenska model).
- Identify and define achievable shared objectives
- Identify sources of funding within the state and voluntary sectors
- Secure consensus and buy-in to a joint proposal/ joint an action plan

Actions:

- Establish stakeholders and core partners willing to participate actively M1 -M3
- Set up one or more discussion meetings to take views, secure commitments, formulate a directed policy, and review funding opportunities M 3-6
- Investigate Agrenska model – how it works, benefits, agencies involved, would it work in UK? M2
- Establish funding opportunities M6
- Establish costed programme M6
- Publish prospectus M 7

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