# Initial ideas for review of psychosocial support for those living with Alport Syndrome – *review dedicated to the memory of Anthony Whitworth*

*Draft 7 May 2014*

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## Why a review of psychosocial support?

Alport Syndrome is an inherited disease that causes kidney failure, hearing loss and eye abnormalities. It can impact a number of individuals in a family and individuals 'live with it' throughout their lifetime. With kidney transplants available and access to proper hearing aids and support, there is lots of potential for people living with Alport Syndrome to live life to the full. There are a number of individuals who have interesting lives or careers or who do inspirational things in their lives; their stories and how they cope with specific challenges will inspire future generations and are worth sharing. Alport Syndrome impacts different people in different ways throughout their lives and there are also some key stages or 'crunch points' where recognisable challenges are faced. We need to understand what are these key stages and the specific challenges and what needs to be in a 'support toolkit' for individuals and families to help them aim towards a brighter future.

## Scope

- Recommendations will draw upon a core 'toolkit' of solutions available to all. The 'toolkit' may have elements that are 'self-help' solutions' or services that are available through the NHS or related services, and the 'toolkit' will also tap into alternative forms of support or therapies available through other recognised channels.
- As the key stages and 'crunch points' are better understood, these will be looked at and compared with the key stages of a 'normal' persons' journey through life to better understand the cumulative impact of dealing with normal life plus a life-long condition on top of it.
- Wherever possible we want this to be a positive enquiry and to celebrate and share good stories and experiences, but it is also important to understand the experiences of those who do not have such positive stories and what would have helped in their situations.
- This review will provide recommendations for those living in the UK, but will need to draw upon relevant international work. The recommendations may also be relevant for other English speaking countries around the world.
- Although our focus is a review for Alport Syndrome patients, carers and families, there is much to be gained by both learning from and sharing with other inherited rare disease groups or other related areas eg renal, transplantation, deafness etc. where there may already be a lot of experience and solutions to tap into.

## Potential outcomes from this review

*For those living with Alport Syndrome: patients, carers and families*

- A shared understanding of specific needs over time and the key stages where support is needed.
- Information available on the web that helps with diagnosis of issues and ideas for potential solutions.
- Recommendations on how to access or develop elements for a practical 'toolkit' for the future.

*For clinical teams caring for patients with Alport Syndrome*

- A shared understanding of specific needs over time and the key stages where support is needed.

- Shared learning from units that have good support and the value to patients and carers.

*For Alport UK*

- Material to share on the web as a 'toolkit' with stories, resources – stories of challenges and how to solve them.
- Guidance on what Alport UK needs to do, to lobby for or put in place to provide more effective support for those living with Alport Syndrome.
- Recommendations on any shared services required to support people.

*For those living with related inherited rare diseases*

- An insight into what needs are shared with others and input to the recommendations.
- Opportunity to collaborate on solutions – shared numbers of patients will mean that the ideas are taken more seriously.

## Examples of key stages or 'crunch points' where specific help is needed

- **Hearing loss -** From a recent survey (December 2013) of those living with Alport Syndrome, the most important issue impacting quality of life was 'hearing loss'. There must be tools and experience to tap into from Action for Hearing Loss etc. The issues are compounded if the individuals also are dealing with eye issues.
- **Diagnosis** – this can be a shock for anyone, particularly if there is little credible information available and there are key decisions to be made quickly in terms of treatment etc.
- **Diagnosis of children** - Parents anxious about implications for their impacted children.
- **Decisions around family planning** – before, during and after decisions made, particularly as family planning is a very personal and private subject and there are many choices to consider with very different outcomes.
- **Teenagers** – needing extra support as they learn about the science of the body in their school curriculum and begin to realise the implications for their future at a time where their brains, bodies and emotions are experiencing major change anyway and dealing with the pressures to perform in key external exams such as GCSEs, A-Levels and undergraduate studies etc.
- **Young adults** – coping with declining health and the 'normal' challenges of further education, moving onto full-time work and relationships.
- **Carers and families** watching their loved ones' hearing or health declining.
- **Sibling 'guilt'** of not being impacted or of siblings impacted 'getting all the attention'.
- **Parental feeling of 'guilt'** having passed on the gene to their children.
- **Loneliness for adults**, men in particular (as recent studies showed that men in their early 40s are the most vulnerable group prone to mental illness at this stage), dealing with deafness, eye or renal complications etc on top of usual challenges of life and relationships.
- **Loneliness for carers**, women in particular, supporting partners or children through dialysis, transplantation, hearing issues etc.

## Potential contributors: relevant research or topics of interest

A list of some existing sources of material to start the review:

- Notes written up from Edinburgh Alport Information Day on what people would like to see in a 'toolkit'.
- Alport Syndrome specific research – anything specifically relevant eg
    - French study
    - Alport Syndrome Foundation, USA
    - Latest on organ donation, lobbying governments – international best practice
    - Hearing
    - Genetic counselling project in Australia.
- Related disease areas:
    - Cystic fibrosis
    - Rare renal diseases or hearing similar situations of hearing loss
    - Inherited diseases with long term implications and particularly impacting children
    - Other general renal disease, renal transplantation or hearing/deafness experience.

## Final review to include

- Documented stories to learn from – both inspirational and those with particular challenges.
- Workshops with those impacted by Alport Syndrome.
- Illustrated journey(s) through the life of people living with Alport Syndrome with clear 'crunch points' identified.
- Recommendations for the elements of a 'toolkit' to be used by those living with Alport Syndrome so they can look at options for support when they are at particular 'crunch points' – this may include self-help, existing services or materials or identify the need for new services specific to Alport Syndrome.

## Practical steps to take this forward

- Get further input to this brief from Whitworth family, experts in elements of psychosocial care.
- Identify/appoint an individual or team to do the review and interviews for us.
- Put a detailed project proposal together that identifies:
    - Who to interview and when
    - Sources to review
    - Timescales and budget.