Describing Alport Syndrome

In the early stages of setting up Alport UK in March 2013, we asked a researcher to interview people living with Alport Syndrome to get their views on:

1. **How would you describe Alport Syndrome very briefly to someone who knows nothing about it?**
2. **What does it feel like to have Alport Syndrome?**

The quotes below are how people responded.

The way people describe Alport Syndrome is quite similar and straightforward. There is no complexity in the way people describe the condition and it is factual, with little emotion, in response to the question:

1 'How would you describe Alport Syndrome very briefly to someone who knows nothing about it?

An inherited kidney disease that leads to kidney failure in some cases and can also affect the hearing

An incurable disease that affects individuals differently, but leads to needing a transplant.

It is a genetic kidney disease that mainly affects boys, and girls are carriers. It affects your hearing around the age of 9 and you go into renal failure in your late teens when you will need a transplant. (This is my family's journey, I do realise other things happen to other families at different ages)

An inherited disease that causes kidney failure and deafness usually in teens/ twenties or later

A rare inherited condition that impacts kidney function and hearing and can cause some eye abnormalities. Those who inherit it will probably require a kidney transplant.

A genetic disease which affects kidneys, often the ears, and occasionally the eyes.

Genetic disease that causes kidney failure and hearing loss, often in multiple family members in each generation.

A genetic disease transmitted from the parents, it affects vision and hearing to various extents

A genetic disease that affects renal function, hearing and eyesight in the young. It can't be prevented, can't be cured and is life-limiting.
I used to describe it as a disease that lies in wait, allowing you to live a perfectly normal life until it one day just pops up and takes away your kidneys!

A deteriorating membrane in the kidney. You have to ultimately change the kidney or compensate because you can't restore the membrane function.

A rare genetic disease involving chronic kidney disease, deafness and eye abnormalities.

A genetic disease which affects the kidneys and eventually leads to kidney failure which can be treated with dialysis and/or transplantation.

2 However, the question **'What does it feel like to have Alport Syndrome?'** naturally elicits a more emotional response. The reality of living with Alport Syndrome is stated openly and honestly; patients tend to be very straightforward and stoical in their responses; parents express their anxiety more intensely but this is often tempered with mentions of a need for practical steps and responses:

When you first find out it is quite a shock and overwhelming with all this new information but you gradually get used to it. Physically it can make you feel very tired and cause renal morning sickness.

My experience is different to most. I was not educated and, after reaching 18 years of age, believed I wouldn't have to worry about kidney failure. It caught me by surprise. This is why I want to make sure people are educated.

I have had a while to get used to it, so to me it feels normal (sorry I can't provide a better answer here!)

Isolating as with any chronic illness but also devastating because families have to relive the illness in each generation

Our son has Alport's and we find it really hard to talk to him and come to terms with what will happen to him in the future.

Worrying, depressing.

I'm father to a 30 year old girl who was diagnosed at 5 years old. It is the worst thing a parent can hear - that there's nothing that can be done, but wait.. Having to give samples every 6 months etc. You live an anxious life related to the condition. It's like a Damocles sword hanging over you.

The worst thing is to be passive, waiting for samples etc. As soon as you fight, you become more positive. It is important to connect with other people affected by the same disease.

As a female carrier, with an unusual history of the female members (mother, cousin, sister) of our family going into renal failure, it is always at the back of my mind, in everything I do. How can I stay healthy to keep my kidney function and hearing as normal as possible?

But I also watch my sons and know that although they are currently having to deal with the challenges of deafness (sometimes a big challenge on its' own) but will have many more challenges to deal with in the future. That is nerve wracking. If they fall ill, we have to immediately sort what it is to make sure it does not damage their kidneys any further. Our life is about protecting their hearing and kidney function as much as possible for as long as possible, so we can get them through adolescence, schooling and possibly Uni and to an age where they are more able to deal with it themselves,

mentally and physically. If proactively managed, it can help them adjust and live a near- normal life and attend normal schools. Though their ‘normal’ is very different to other people’s ‘normal’ as they deal with deafness and people’s reaction to it.

One of the upsides to it all, is that you become more resilient and able to deal with change, issues and move on and make the best of any situation. You will also have amazing periods of your life where you can get on normally, work etc once you have had a transplant. One of the downsides is that you have to go through intense periods of hospitalisation, major surgery on many occasions (and not just for a kidney transplant) AND deal with the side effects and juggling of many different drugs which you will have to take for your entire life.

One of the toughest things is that most people have no idea that you have any sort of condition (sometimes this can be a good thing) but when you are feeling rubbish, there are no outward signs (eg hair fallen out, or bandaged arm) so some people assume you are making a fuss of nothing…!