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Founding Members

Who we are and a little about the conditions we represent

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Fiona Copeland

Primary Ciliary Dyskinesia Family Support Group

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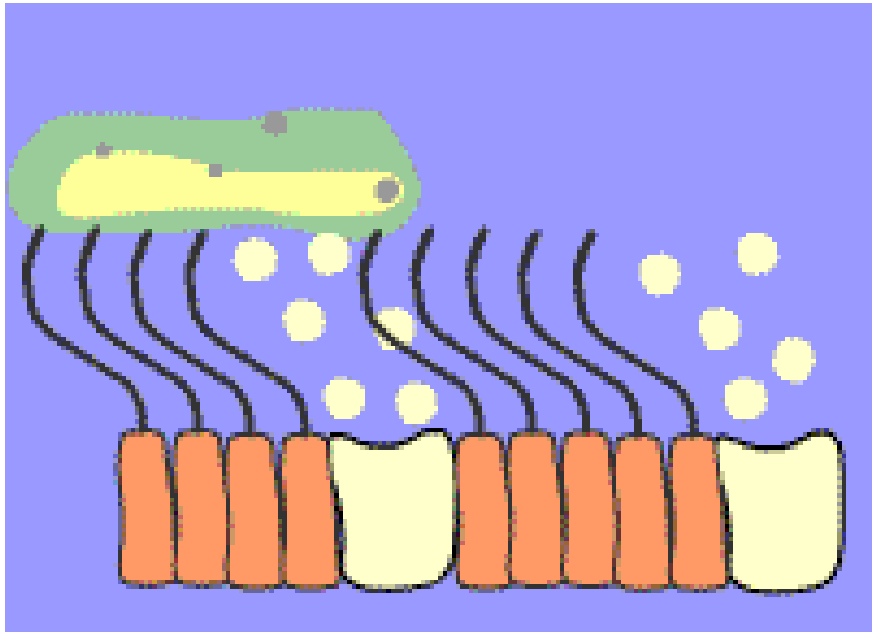
What is Primary Ciliary Dyskinesia?

Normal cilia



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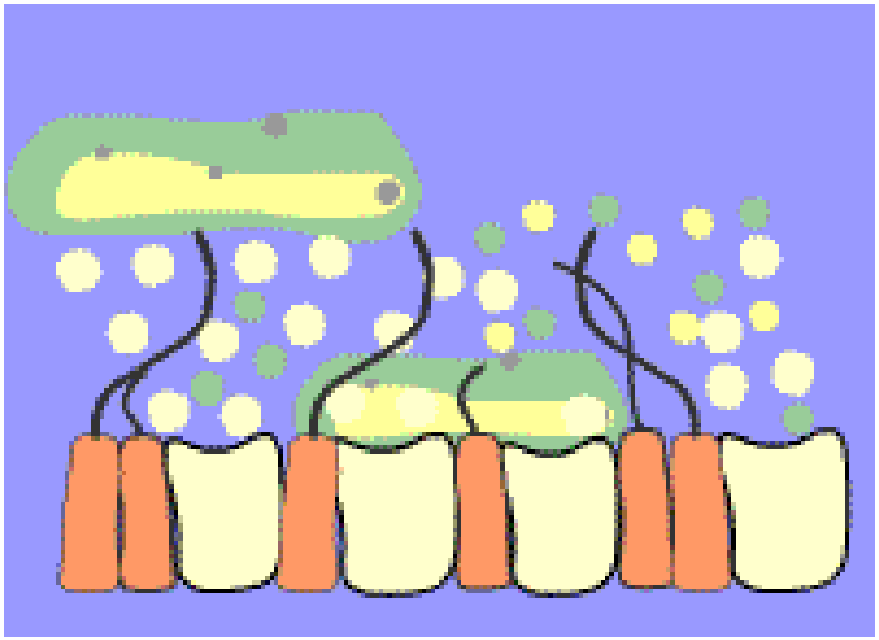
Abnormal Cilia



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How many people are affected?

- PCD- autosomal recessive inheritance

  Incidence 1:15,000

- Higher in the Asian population

  ethnic pockets of disease in areas where consanguinous marriages are prevalent - incidence can be as high as 1:2500

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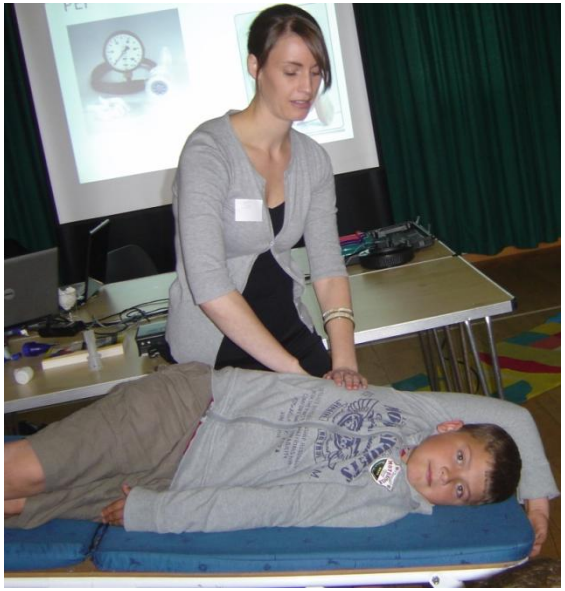
Impact of PCD

- Twice daily physiotherapy
- Targeted antibiotics either orally or intravenously
- Permanent lung damage if not diagnosed
- Hearing problems
- Fertility problems
- Living with a chronic lung condition



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Tess Harris

pkd charity

Polycystic Kidney Disease

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What is PKD?

POLYCYSTIC KIDNEY DISEASE - range of genetic diseases which are a common cause of kidney failure in children and adults. Two main forms:

ADPKD - Autosomal Dominant Polycystic Kidney Disease

ARPKD - Autosomal Recessive Polycystic Kidney Disease

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ADPKD

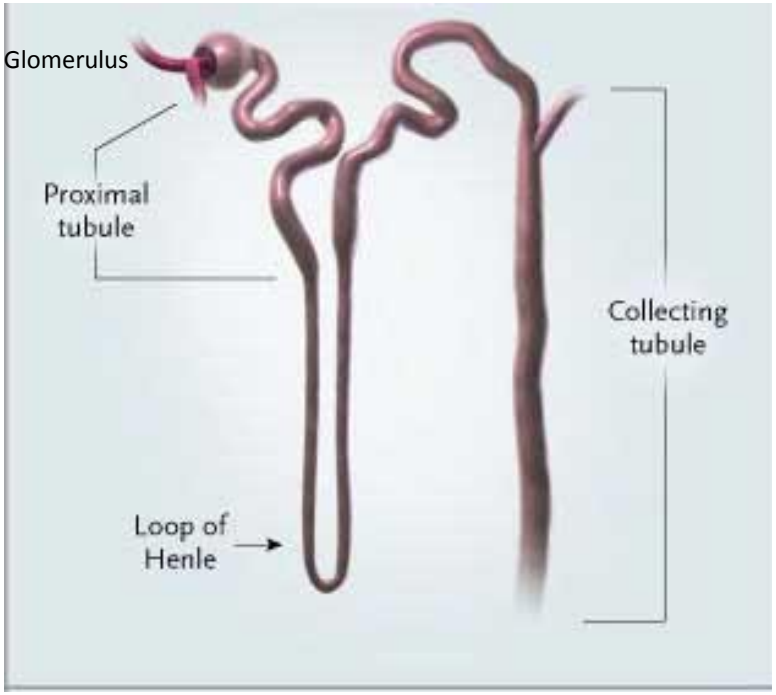
The world's most common inherited life-threatening condition:

- Fluid-filled cysts develop in both kidneys
- Causes progressive renal failure
- Affects other organs - liver, pancreas, spleen, brain, intestines

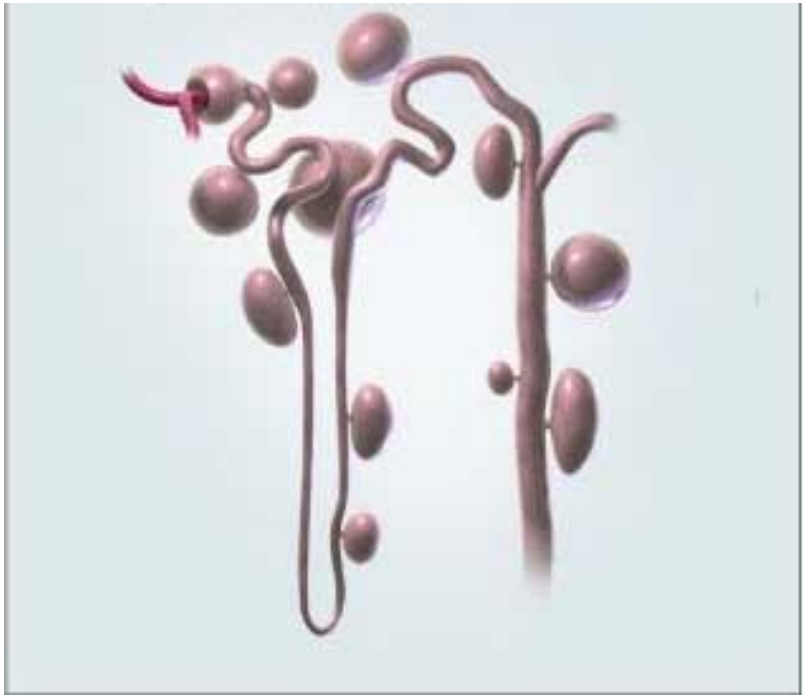
Affects between 1 in 800 and 1 in 1000 – approx 12.5 million worldwide, **70,000+ in UK**. Affects men and women equally with no apparent racial bias.

Two genes: PKD1 and PKD2 with differing outcomes. Typical age of onset: 30s to 50s.

Normal kidney nephron



ADPKD kidney nephron



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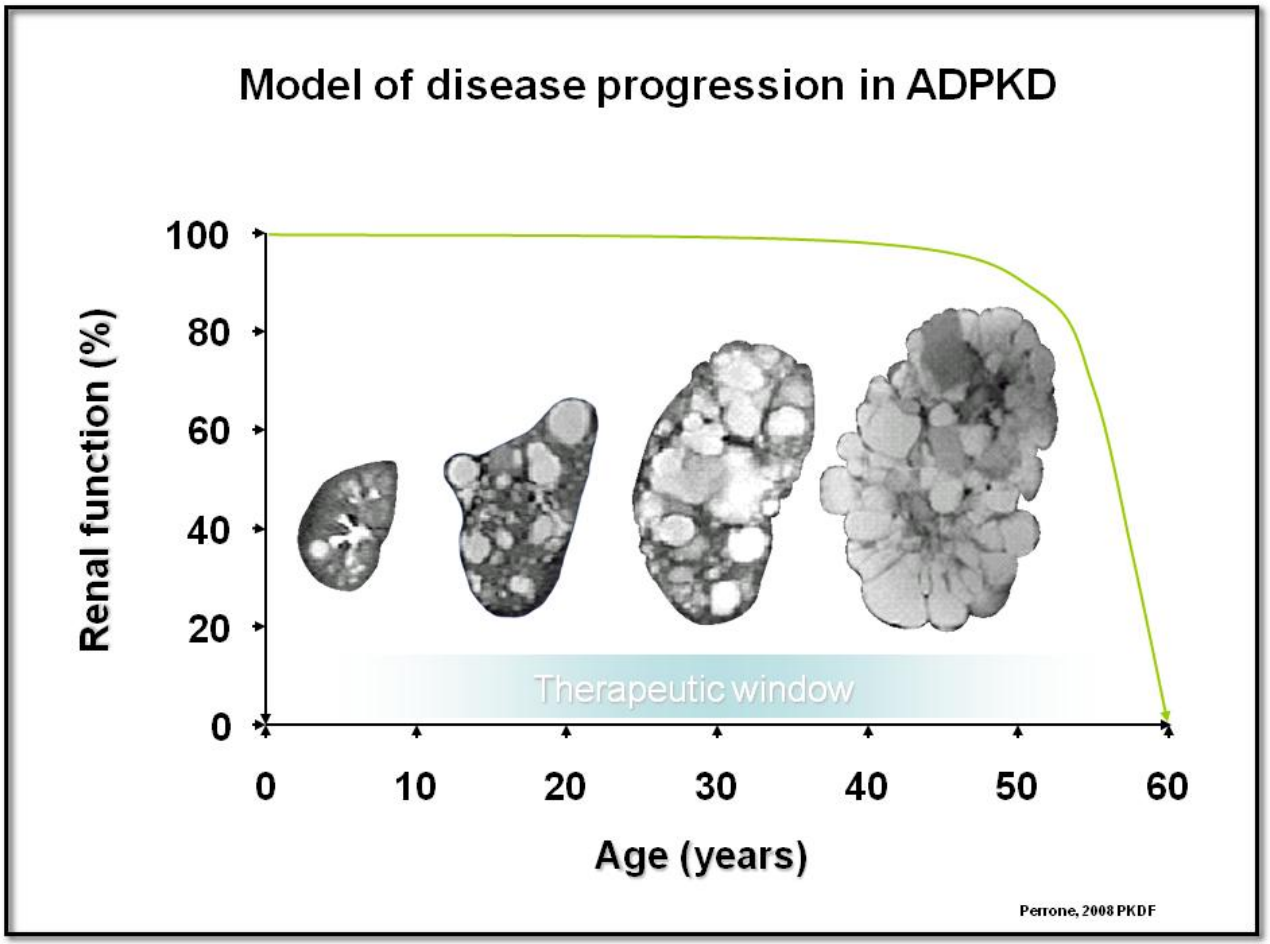
Impact of ADPKD



PKD1
End-stage RF: 53 yrs

PKD2
ERF: 73 yrs

Highly **variable** even in families

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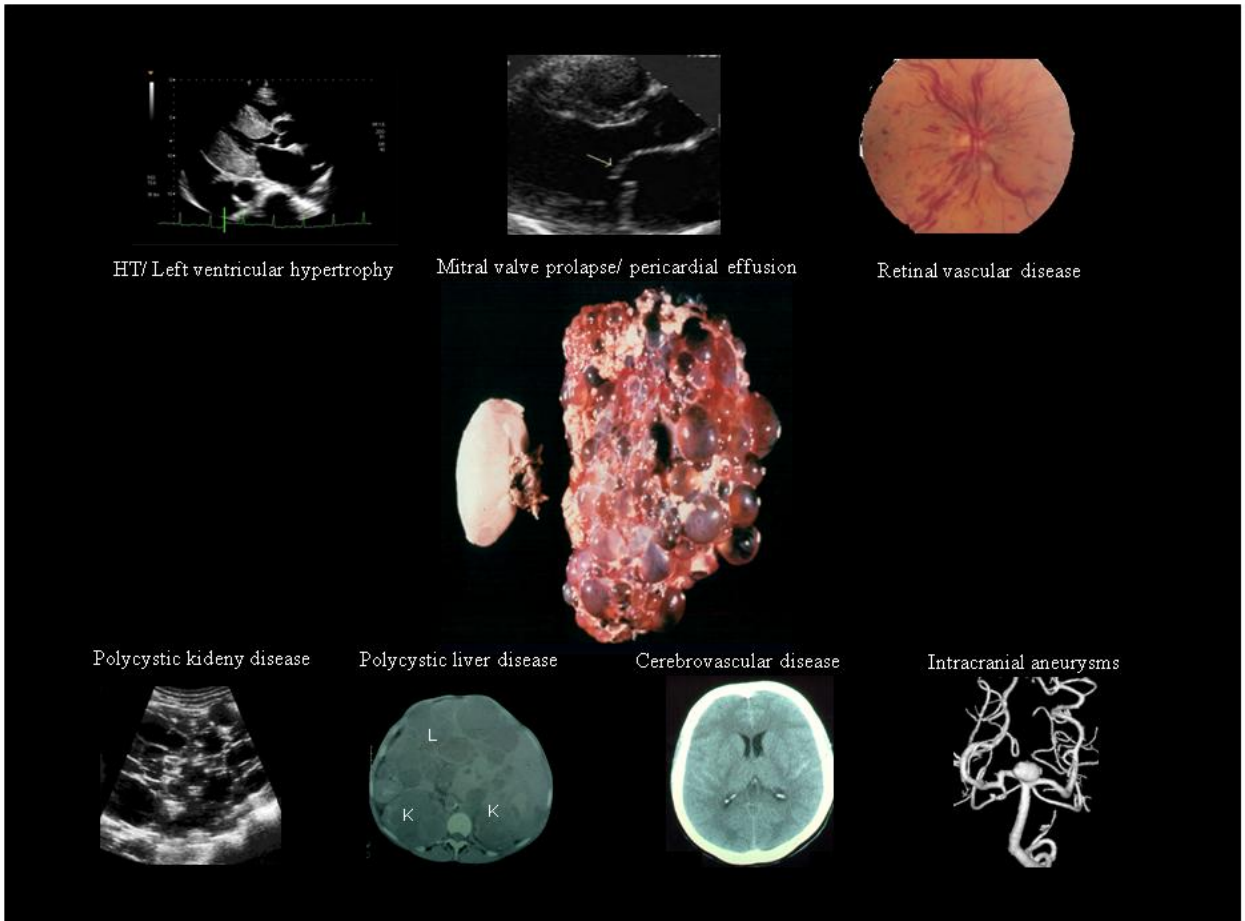
Impact of ADPKD



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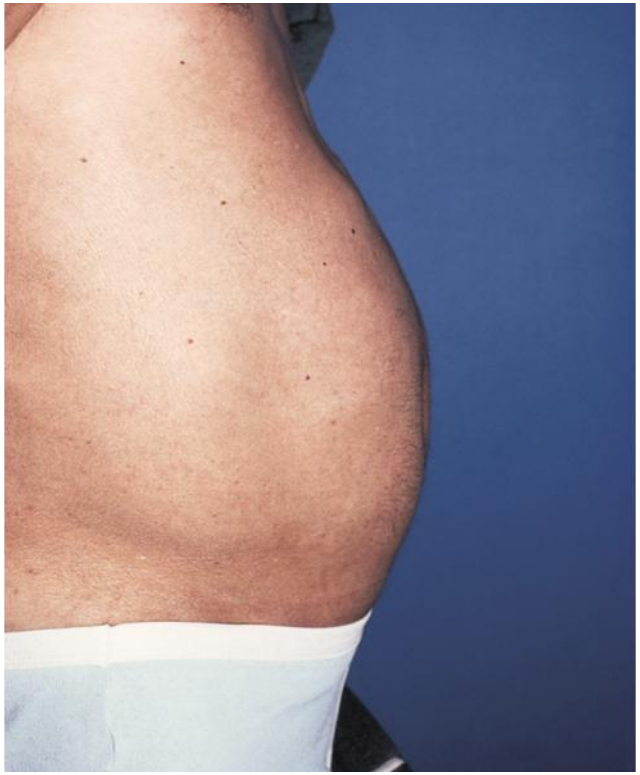
Impact of ADPKD



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ARPKD

1 in 20,000 live births - **3000+ in UK**. Higher incidence in Finnish and Afrikaaners.

Causes enlarged kidneys with or without cysts, liver enlargement and often high blood pressure.

Typical age of onset: newborn and often presents in utero.

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Impact of ARPKD

In utero – sometimes fatal

- Enlarged kidneys and little amniotic fluid
- Failure of the lungs to fully develop
- Deformities of spine and limbs

Newborn - 30%-50% die at birth or shortly thereafter

- Respiratory failure needing ventilation in ~40%
- 12% of these children develop chronic lung disease
- May require a nephrectomy
- Problems with salt and water balance, hypertension
- Poor kidney function, infections

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Impact of ARPKD

With age

- Hypertension
- Kidney failure often by age 30
- Liver abnormalities, primarily Congenital Hepatic Fibrosis

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Tonia Hymers

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What is LMBBS?

Laurence-Moon-Bardet-Biedl Syndrome is a rare, recessively inherited disorder, which commonly displays these symptoms:

- Visual impairment, often leading to blindness
- Obesity
- Extra fingers and/or toes (polydactyly)
- Developmental delay, speech and co-ordination problems, and often, learning difficulties
- Kidney abnormalities, often leading to transplant
- Hypogonadism is common amongst affected males
- Other health problems may occur

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How many people are affected?

- In the UK, approximately 1 in 150,000 babies are born with LMBBS
- Some Asian communities have higher incidences owing to consanguineous marriages.
- At present, 280 adults and children are known to the LMBB Society.

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Impact of LMBBS

- Complex syndrome, delayed diagnosis is common.
- No treatment for rod-cone dystrophy , correct early diagnosis vital for future development.
- Obesity difficult to treat, lifelong commitment to healthy diet and exercise necessary.
- Extra digits surgically removed at young age, with successful results.
- Learning difficulties and speech problems need early intervention for successful outcome.

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Impact of LMBBS

- Kidney problems can usually be treated, severe cases require transplantation.
- Dietary advice and/or tablets can treat hypertension. Certain heart defects may be correctable.
- Reproductive system defects, hormonal and fertility problems may require treatment.
- Medical intervention and monitoring becomes a way of life for those affected by LMBBS

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Kerry Leeson-Beevers

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What is Alström Syndrome?

Alström Syndrome is characterised principally by a number of key conditions:

- Retinal degeneration (Rod Cone Dystrophy, Nystagmus and Photophobia)
- Sensorineural hearing loss (ranging from a mild, moderate to severe loss)
- Obesity
- Insulin resistance

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What is Alström Syndrome?

Additional features can include:

- Cardiomyopathy (poor cardiac function where the heart muscle is weakened and enlarged)
- Type 2 diabetes
- Renal and hepatic dysfunction (affecting the kidneys and liver)
- Hypertriglyceridaemia and tryglycerides (elevation of fatty substances found in the bloodstream)

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How many people are affected?

- 47 families known to AS UK

- Rising number of Asian families - ethnic pockets of disease in areas where consanguineous marriages are prevalent

- 400 children and adults in the UK could be suffering from Alström Syndrome

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Impact of Alström Syndrome

- Multi-disciplinary care is needed
- Dual Sensory Loss is frequent
- Constant requirement to manage diet, exercise and drug regimes
- Complex and progressive disease
- Very little knowledge and awareness of Alström Syndrome
- Isolation – Alström Syndrome is a very rare condition

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Impact of Alström Syndrome

- Our young people attend a variety of different educational settings but they must be supported by professionals who have an understanding of visual and hearing impairment

- Direct Payments and Individualised Budgets have proven to be an effective way to support people to lead an independent, healthy and active life

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Summary

- ASUK initiated the worlds first multi-disciplinary clinics for the condition
- ASUK maintain a National database
- ASUK provide family liaison support
- ASUK initiate research
- ASUK provide the link between children and adult services
- ASUK developed and maintain the only web site www.alstrom.org.uk on the condition.
- ASUK designed the only information leaflet and medical handbook available on the condition.
- ASUK are a founder member of the CA UK

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Alström Syndrome UK and the Ciliopathy Alliance

- Able to identify areas of common concern- e.g. PCD
- Better prospects for research
- Consultants have wider access to expertise
- Being very rare – joining with others makes us more visible
- ASUK strongly supports CAUK

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How to get involved

- Find out more about our support groups by visiting our stands today
- Join the CAUK today
- Visit our website www.ciliopathyalliance.org.uk and sign up to our mailing list
- Or email info@ciliopathyalliance.org.uk
- Spread the word to patients and colleagues