



# Founding Members

Who we are and a little about the conditions we represent

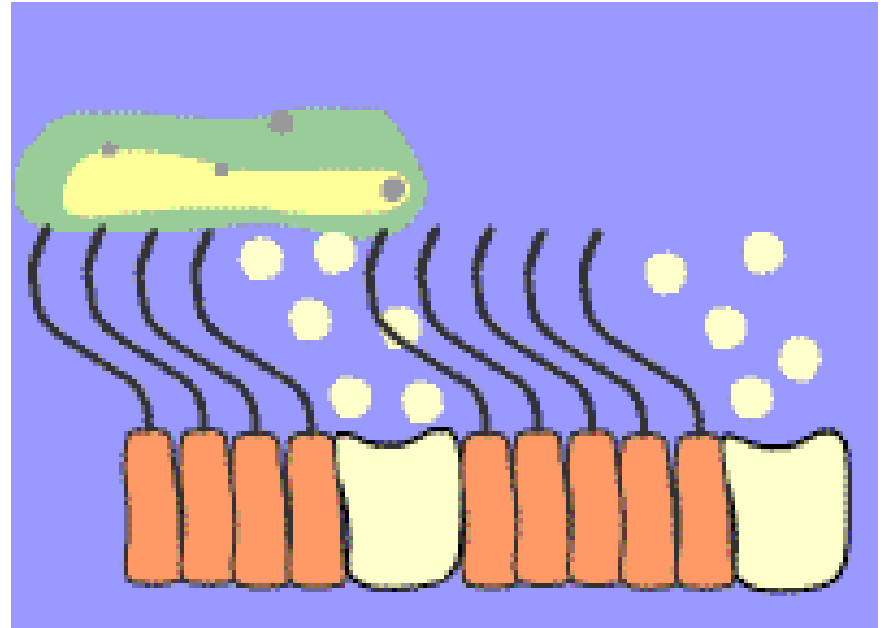


# Fiona Copeland

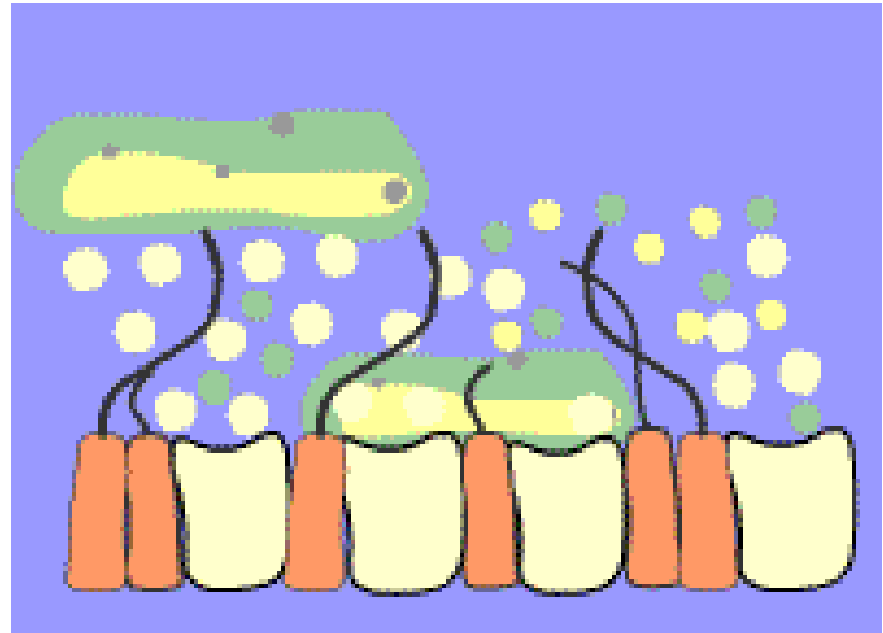


# What is Primary Ciliary Dyskinesia?

Normal cilia



# Abnormal Cilia



# 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

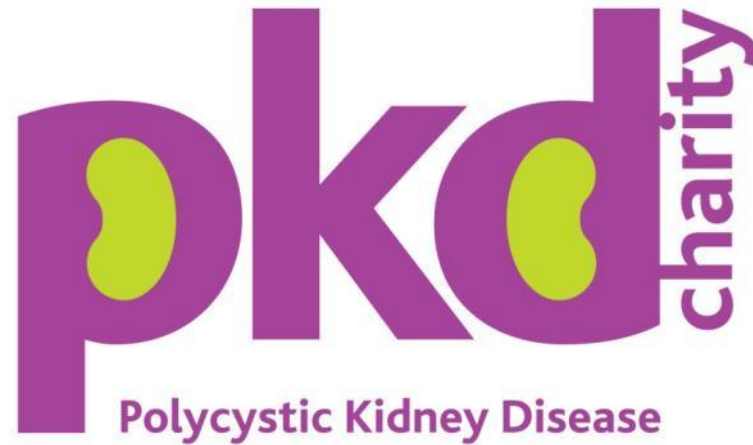
# Impact of PCD

- 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





Tess Harris





# 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





# ADPKD

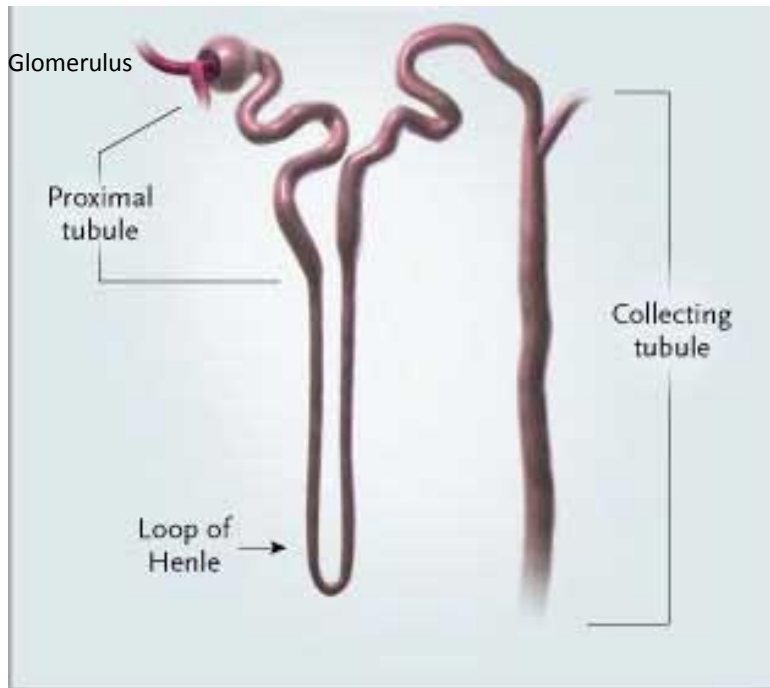
The world's most common inherited life-threatening condition:

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

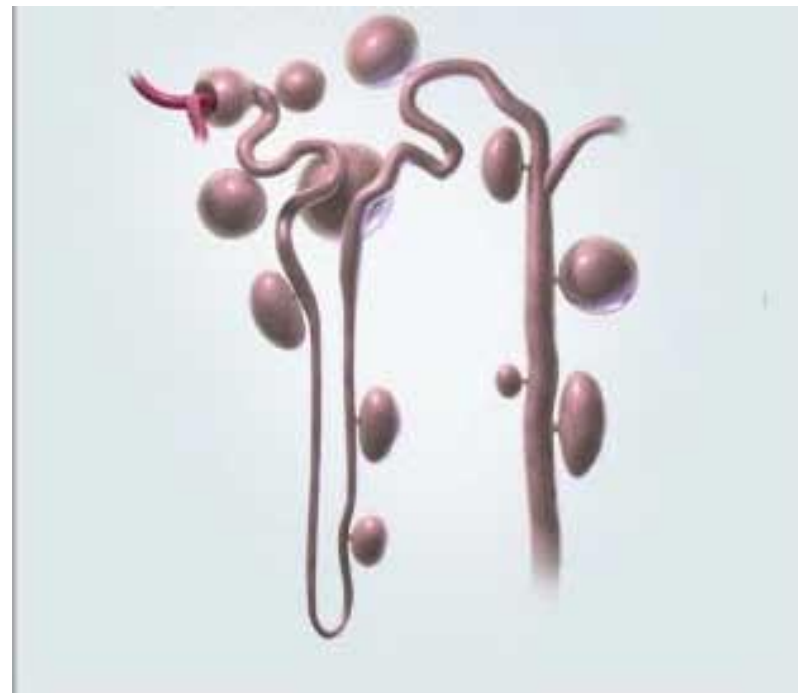
Affects between 1 in 800 and 1 in 1000 – approx 12.5 million worldwide, **70,000+ in UK**. Affects men and women equally with no apparent racial bias.

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

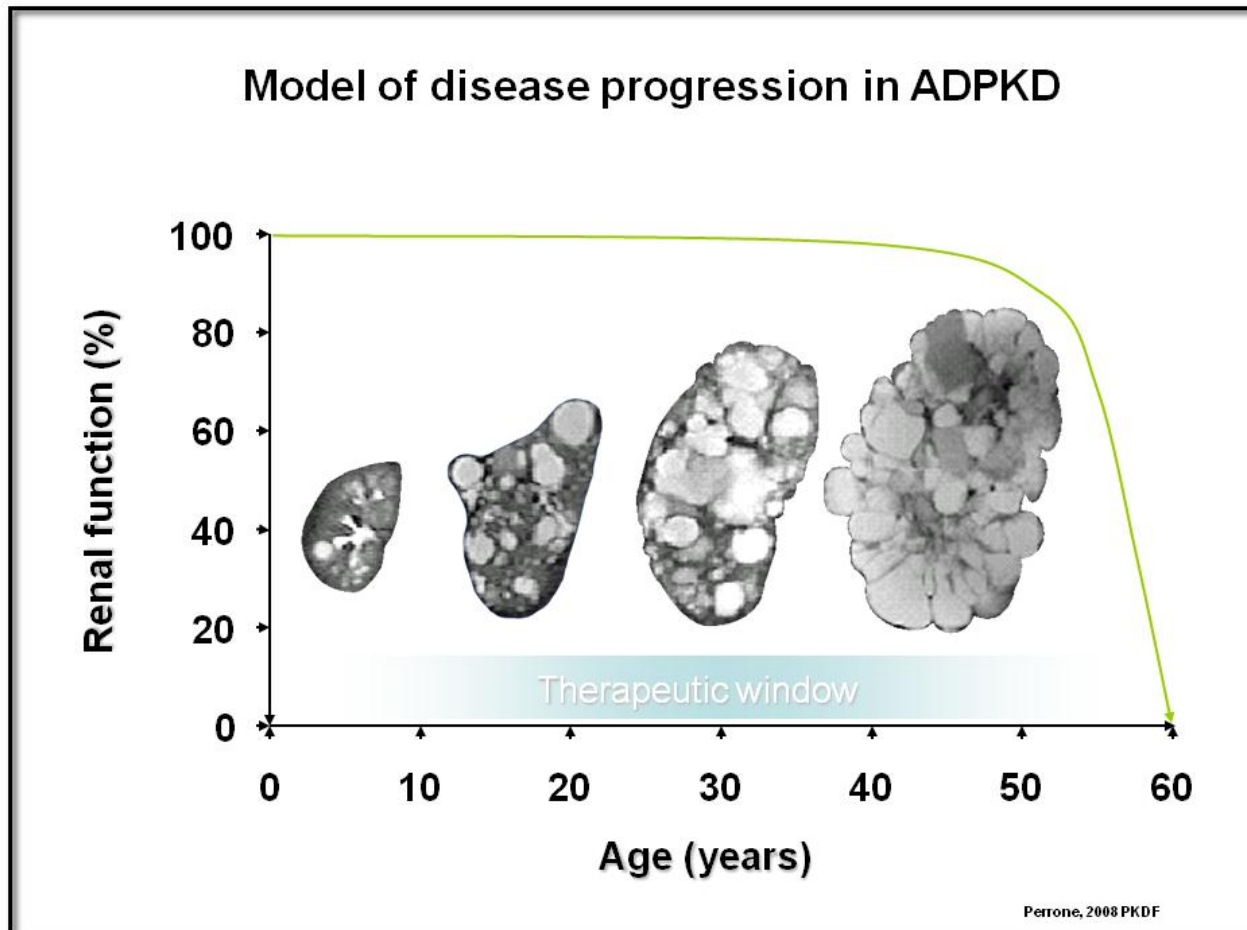
Normal kidney nephron



ADPKD kidney nephron



# Impact of ADPKD

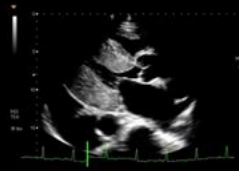


**PKD1**  
End-  
stage RF:  
53 yrs

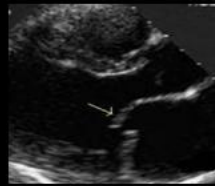
**PKD2**  
ERF: 73  
yrs

Highly  
**variable**  
even in  
families

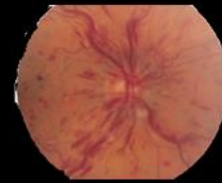
# Impact of ADPKD



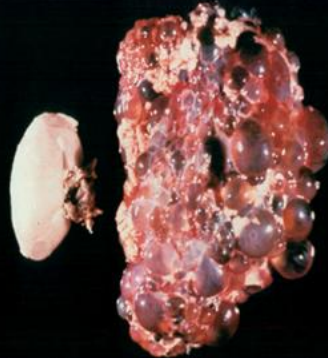
HT/ Left ventricular hypertrophy



Mitral valve prolapse/ pericardial effusion



Retinal vascular disease

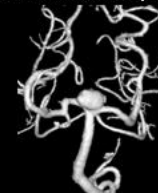
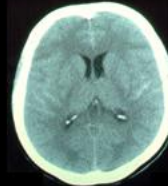
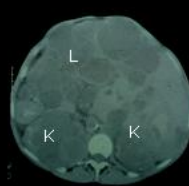


Polycystic kidney disease

Polycystic liver disease

Cerebrovascular disease

Intracranial aneurysms



# Impact of ADPKD





# 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.

# 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



# Impact of ARPKD

## **With age**

Hypertension

Kidney failure often by age 30

Liver abnormalities, primarily Congenital Hepatic Fibrosis





Tonia Hymers



# 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

# 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.

# 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.

## 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



# Kerry Leeson-Beevers



Alström Syndrome UK  
Support Group

With your help, we have hope

# 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

# 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)





# 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

# 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

# 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



# 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](http://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



# 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



## 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](http://www.ciliopathyalliance.org.uk) and sign up to our mailing list
- Or email [info@ciliopathyalliance.org.uk](mailto:info@ciliopathyalliance.org.uk)
- Spread the word to patients and colleagues