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## **History in Nephrology**

*Legends whose contributions inspire us even today!*

*Alport Spectrum Disorder*



***Arthur Cecil Alport***

## ***Arthur Cecil Alport***

**Was a British Physician who first clearly described the hereditary association of**

- **Kidney Disease**
- **Hearing loss**
- **And amilial occurrence**

**In 1927**

**He published observations of a family in which several members had progressive nephritis and deafness, recognizing that the condition was inherited.**

**AT that time, the condition was called:**

- Hereditary familial nephritis**
- Later popularly termed Alport Syndrome**

## ***Modern Terminology***

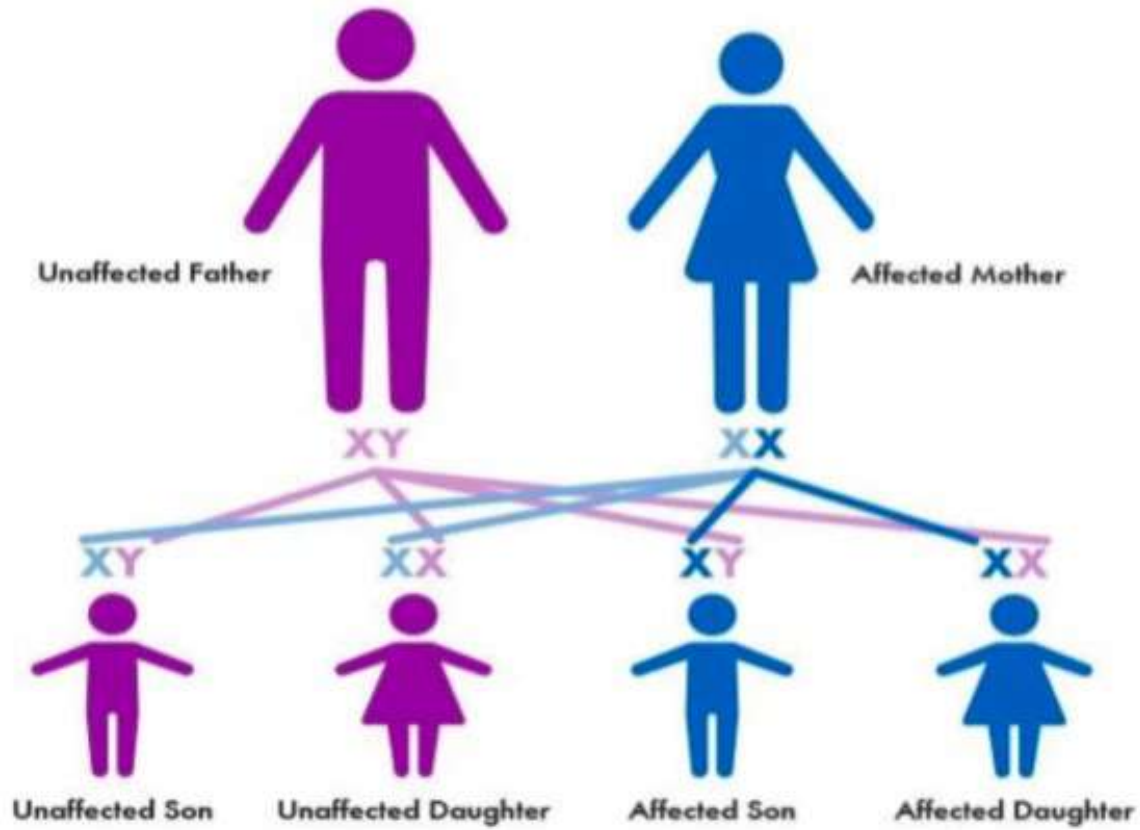
Today many experts use broader terms such as:

- **Alport spectrum disorder**
- **COL4A – associated nephropathy**
- **Type IV collagen nephropathy**

This reflects that the disease ranges from:

- **Mild isolated haematuria to**
- **Severe early kidney failure with deafness and ocular disease**

**XLAS**  
**X-Linked Alport Syndrome**  
Affected Mother



**X** is the gene from the Mother that carries Alport Syndrome. Each child will have a 50% probability of inheriting the Alport Syndrome gene from the affected Mother.



# UNDERSTANDING THE ALPORT SPECTRUM

## One Gene, Many Faces

### A Spectrum of Type IV Collagen Disorders

Different Genes...  
Different Severity...  
Same Spectrum!



#### CLASSIC TRIAD



Kidney Disease



Hearing Loss



Eye Abnormalities

#### MILD

##### Benign Familial Haematuria



Microscopic haematuria only

##### Thin Basement Membrane Nephropathy



Thin GBM  
Often normal kidney function

##### Autosomal Dominant Alport Syndrome



Hematuria ± Proteinuria  
May progress to CKD  
(Usually adult onset)

##### Females With X-Linked Alport Syndrome



(one affected)

Variable severity  
Hematuria ± Proteinuria  
Risk of CKD

##### Alport Syndrome (Mild Genotype)



Early hematuria  
Progressive CKD  
Later onset

##### Alport Syndrome (Severe Genotype)



Early onset  
Proteinuria  
Progressive CKD  
Kidney failure

#### SEVERE

Genes commonly involved: COL4A3, COL4A4, COL4A5, FN1

#### KEY TAKEAWAYS

- Think Alport in any patient with persistent haematuria
- Early diagnosis helps slow progression
- Genetic testing guides management & family screening
- ACEi/ARB, BP control & early care make a difference

#### WHAT IS ALPORT SPECTRUM?

Alport Spectrum Disease includes a range of kidney disorders caused by mutations in type IV collagen genes. It ranges from isolated haematuria to kidney failure, often with hearing loss and eye abnormalities.



Recognize early, Treat early, Change outcomes.

#### INVITATION TO DISCUSSION (NOT WEBINAR)

We cordially invite you to an exclusive discussion for Nephrologists

### "ALPORT SPECTRUM DISEASE: FROM GENE TO CLINICAL CARE"

Date: Monday 11/5/2026

Time: 09:00 - 10:00 PM IST

Platform: Zoom Meeting

Join Us to Explore the Spectrum, Improve Outcomes!



LEARN • SHARE • ADVANCE  
Together for Better Kidney Care



3 QUIZES  
3 PRIZES!



QUIZ 1  
PRIZE 1



QUIZ 2  
PRIZE 2



QUIZ 3  
PRIZE 3

Be a Part of the Knowledge.  
Be a Part of the Change.

