



# ALSTROM SYNDROME- AN INTERESTING AND RARE CAUSE OF CKD

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## Introduction

- Alstrom syndrome is a rare genetic multisystem disorder.
- It is an autosomal recessive disorder resulting from mutations in the ALMS1 gene.
- Disease manifests in infancy characterised by vision and hearing loss, cardiomyopathy, diabetes mellitus
- Progressive kidney disease occurs in childhood leading to end stage renal disease.

## Case Description

- 24 year old female with childhood history of progressive vision and hearing loss from 2 years of age
- Presented with fracture of right femur to Orthopedics OPD.
- There was history of consanguinity amongst the parents.

- Blood investigations revealed renal failure. She was referred to nephrology.
- Urine routine showed no proteinuria.
- No history of childhood UTI, OTC drugs.
- Imaging showed bilateral contracted kidneys
- Mineral bone profile showed Vitamin D deficiency and hyperparathyroidism which was attributed for fracture.
- Further investigations revealed diabetes mellitus and hypothyroidism.
- Cardiac evaluation revealed dilated cardiomyopathy with portal hypertension.
- With high suspicion of multisystem genetic disorder, genetic analysis was done which revealed mutation in ALMS 1 gene.
- Diagnosis of Alstrom disease was confirmed.
- She has good urine output.
- She is currently in stage 4 chronic kidney disease and is on followup with us.

## Conclusion

- Alstrom syndrome is a rare genetic disorder.
- This disorder is generally characterised by vision and hearing abnormalities, obesity in childhood, insulin resistance, diabetes mellitus, dilated cardiomyopathy and progressive renal dysfunction.
- Intelligence is usually unaffected.
- Mutations in ALMS1 gene has been implicated in ciliary function, cell cycle control and intracellular transport.
- It should be considered for childhood chronic disease with visual and hearing abnormalities.
- Genetic analysis confirms the diagnosis.
- Management is conservative.
- Depending on the other system involvement, renal transplantation can be considered as a choice.