



## Scenario of MARFANS Syndrome in a Kashmiri Family

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**Received:** October 10, 2023

**Published:** November 01, 2023

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Marfans Syndrome Is not an uncommon condition kind of connective Tissue Disorder which involves galaxy of tissues like bones ligaments joints blood vessels hear valves skin chest skeletal muscular system eyes heart etc. this syndrome complex is characterized by

- Skeltomuscular Manifestations
- Occular Manifestations
- Cvs manifestations

Tall person tower skull macroglossia high arched palate Macroglossia Long Tapering spidery fingers Arachnadectaky kyohoses scoliosis span of arm is greater than height.

Sublaxatiin and dislocation of lens sheprophakia ECTOPIA LENTES nystagmus squint keratoconus cataract glaucoma.

Aortic dilatation mitral valve prolapse Syndrome severe mitral regurgitation pda ATRIAL SEPTAL defects.

As we have already taxed marfans Syndrome being a complex of

- Skeltomuscular manifestations
- Occular manifestations
- Cvs manifestations

Skeltomuscular manifestations are tall person with Tower skull macroglossia high arched palate Long tapering spidery fingers Span of arm is greater than height metacarpasal index is positive Arachnadectaky kyohoses SCOLIASIS.

Pigeon shaped chest and Tendency for Features.

Occular manifestations are

Sublaxatiin and dislocation of lens sheprophakia ectopia lentes nystagmus squint kerataconus cataract glucoma High myopia and Retinal detachment.

Cvs manifestations are

- Aortic dilatation
- Mitral valve proplase Syndrome
- Severe mitral regurgitation
- PDA
- Atrial septal Defects

### Materials and Methods

- I saw a family of MARFANS Syndrome in my office recently in 2023
- 59 years old patient tall with tower skull macroglossia high arched palate Macroglossia Long Tapering spidery fingers metacarpasal index was positive pigeon shaped chest
- Had Bilateral sublaxatiin of lens
- RE was amblyopic
- Left eye was operated for cataract with post fixated iol with a satisfactory vision of 6/18
- He had no cvs manifestations of MARFANS
- His elder son accompanying father was also marfaniod with Bilateral sublaxatiin of lens and his R eye operated for cataract with post fixated SCLERAL iol with good vision
- No cvs manifestations of marfans
- Younger daughter present with parents was normal.

### Results and Discussion

Marfans Syndrome is a connective Tissue Disorder a syndrome complex characterized by

- Skeltomuscular manifestations
- Occular manifestations
- And Cvs manifestations

However we may not find all the 3 manifestations

- So It is very important to rule out
- The all the manifestation
- Be it skeltomuscular
- Occular and Cvs manifestations.

### Acknowledgement

Nil.

### Conflict of Interest

Nil.