



Brief Scenario of Marfan Syndrome

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Marfan syndrome is a kind of connective tissue disorder which is characterized by loss of elasticity basically the disorder involves cartilage bones tendons skin blood vessels CNS lungs heart valves it is a kind of genetic disorder which involves both males and females equally its prevalence in USA is 1 in 5000 both sexes and both races.

Marfan syndrome is characterized by

- Ocular manifestations
- Skeletomuscular manifestations
- C V S manifestations

Ocular manifestations are subluxation and dislocation of lens spherophakia ectopia lentis keratoconus nystagmus squint cataract high myopia retinal detachment.

Skeletomuscular muscular manifestations are tall thin built person tower skull high arched palate macroglossia long tapering spidery fingers kyphosis scoliosis pigeon shaped chest tendency for fractures crowded teeth flat feet arachnodactyly span of arms is greater than height.

CVS manifestations are

- Aortic dilatation
- Mitral valve prolapse syndrome
- Severe mitral regurgitation

- Cong heart disease
- P DA
- Atrial septal defects.

DX by

- Genetic studied
- Chest x-ray
- ECG
- Transesophageal ECG
- ECHO
- CT scan brain
- MRI scan brain.

Treatment by

- Genetics
- Ophthalmologist
- Cardiologist
- General surgeon
- Dentist
- Orthopaedic specialist.

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