EDS ECHO SUMMIT SERIES

PRESENTATION

Myopathic Ehlers-Danlos syndrome (mEDS)

SPEAKER

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Nothing to disclose

What is myopathic Ehlers-Danlos syndrome?

- Myopathic EDS (mEDS) is a rare type of EDS.
- mEDS was first reported in 2014.
- 19 individuals with a genetically confirmed diagnosis of mEDS are known in the medical literature.

What are major features of mEDS?

- Muscle weakness and/or muscle wasting that is present from birth, tends to get better with age, with some worsening during forties.

- Movement limitation of knee, hip and elbow joints due to stiffening/tightening of connective tissue.

- Hypermobility or flexibility of finger/toe joints.

What are minor features of mEDS?

- Soft, doughy skin
- Atrophic scarring
- Motor developmental delay
- Myopathy on muscle biopsy

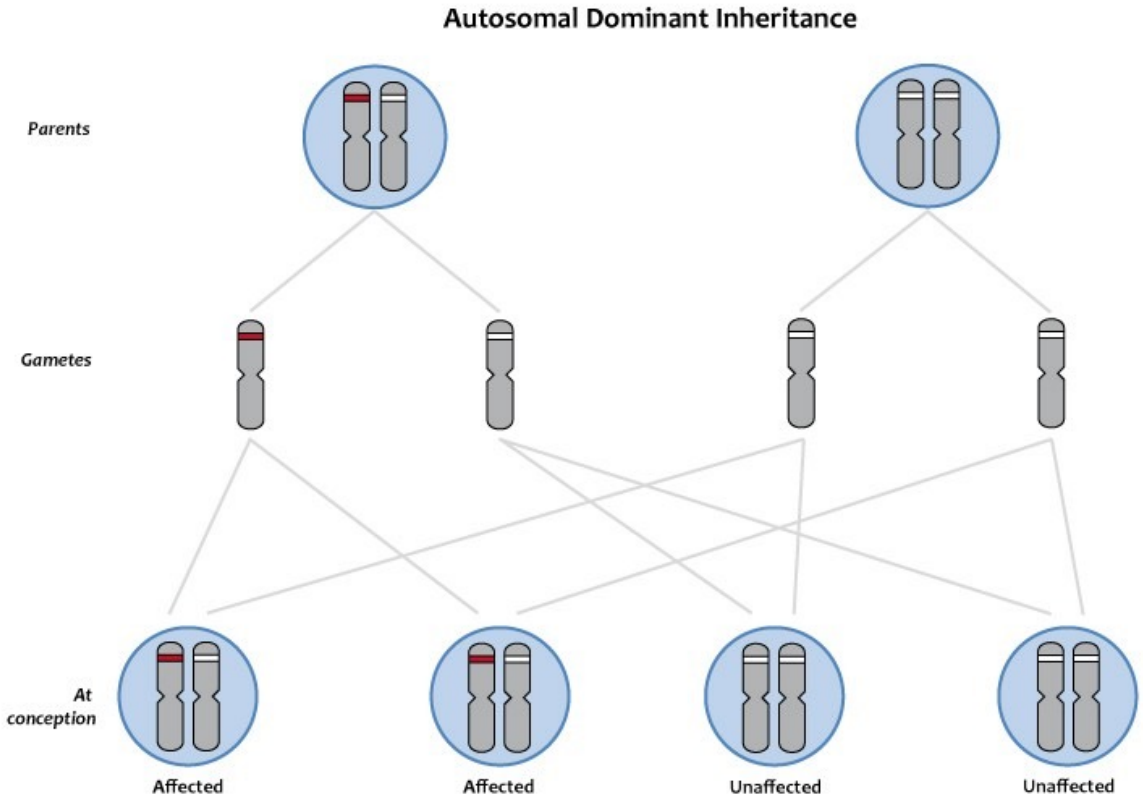
What is the genetic cause of mEDS

- mEDS is caused by alterations in the gene *COL12A1* which encodes the protein collagen type XII.

- In the human body, collagen type XII is among others found in the ligaments, skin, and skeletal muscle.

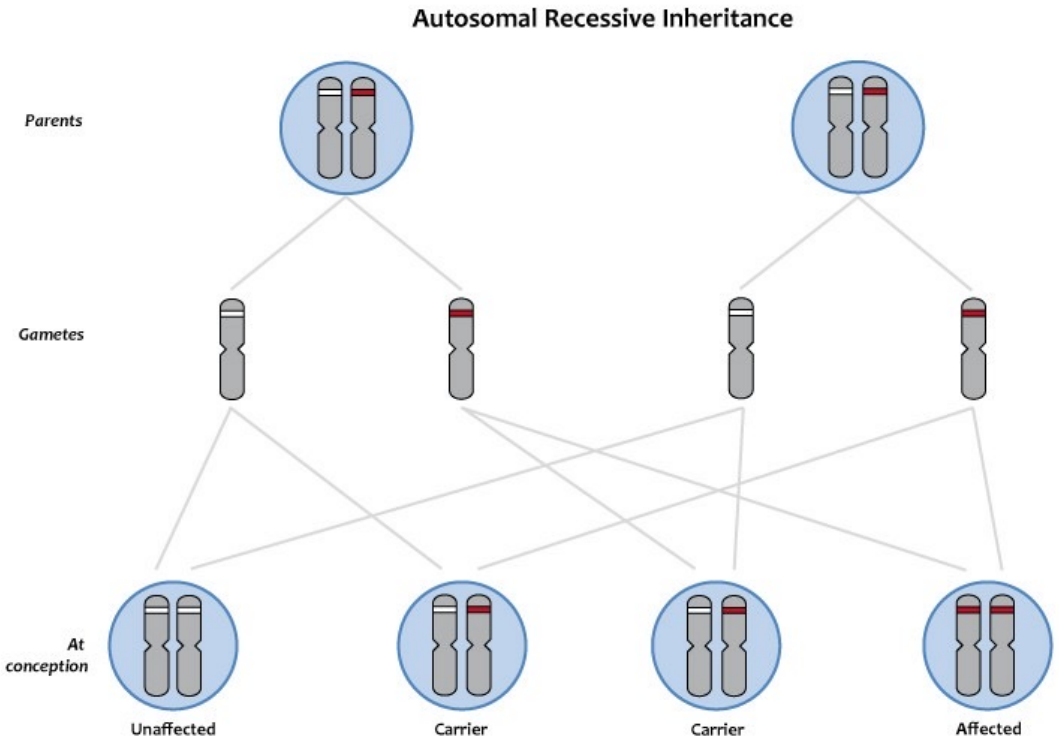
How is mEDS inherited?

COL12A1 gene alterations are mostly inherited in an autosomal dominant way (n=17 individuals)



How is mEDS inherited?

COL12A1 gene alterations are rarely inherited in an autosomal recessive way (n=2 individuals)



Can we predict severity of clinical features based on genetic cause?

- Recessive *COL12A1* alterations that lead to *complete* absence of collagen type XII production cause severe clinical features

- Dominant *COL12A1* alterations that lead to *partially abnormal* production of collagen type XII cause variable, moderately severe clinical features

Which conditions resemble mEDS?

- Genetic conditions characterised by muscle weakness from birth due to disease causing gene alterations in the genes *COL6A1/2/3* encoding collagen type VI.

- These conditions are named Bethlem myopathy and Ullrich myopathy.

What is the management of mEDS?

- No specific guidance for people with mEDS exists.
- Often neuromuscular specialist, occupational and physical therapist are involved.
- Management should focus on preventing complications and improving the presenting symptoms.
  - physical/occupational therapy for stiffening/tightening of connective tissue and;
  - Monitoring feeding/respiratory difficulties.

Is there guidance for pregnant women with mEDS?

- There is no specific guidance during pregnancy.

- Based on guidance for related conditions:

  - lung function surveillance
  - Pre-delivery assessment hip dislocation/contractures
  - Assessment of the overall level of muscle weakness

How to improve care for people with mEDS?

- Natural history of additional individuals with genetically confirmed mEDS
- Further laboratory investigations into function of the protein/gene therapy etc.
- This will help development of guidelines for management and potentially therapy in individuals with mEDS.

Thank you for your attention!