

## Complete Information on Desmin related myopathy with Treatment and Prevention

Desmin related myopathies are a heterogeneous group of muscle disorders, morphologically defined by intrasar-coplasmic aggregates of desmin. Desmin is simply expressed in vertebrates, however homologous proteins are established in many organisms. Desmin is also important in muscle cell architecture and structure since it connects many components of the cytoplasm. The sarcomere is a component of muscle cells composed of filaments and myosin motor proteins which allow the cell to contract. When desmin is not functioning properly there is improper mitochondrial distribution, number, morphology and function. Desminopathies are very rare diseases and only 60 patients have been diagnosed with so far, however this number probably does not accurately represent the population due to frequent mis or under diagnosis.

Desmin is one of the earliest protein markers for muscle tissue in embryogenesis as it is detected in the somites of myoblasts. Although it is existing early in the growth of muscle cells it is expressed at reduced levels and increases as the cubicle nears depot differentiation the muscle cubicle matures simply desmin is existing. A similar protein, vimentin, is present in higher amounts during embryogenesis while desmin is present in higher amounts after differentiation. There is some evidence that desmin may also connect the sarcomere to the extracellular matrix through desmosomes which could be important in signalling between the extracellular matrix and the sarcomere which could regulate muscle contraction and movement. Although rare, the possibility of desmin related myopathies should be considered in the presence of bluish rimmed vacuoles on light microscopy and characteristic ultrastructural inclusions.

Common symptoms of the disease are failing and wasting in the distal muscles of the lower limbs which progresses to the hands and weaponry, so to the body, neck and cheek. When the gene for desmin is knocked away it is no longer capable to operate decently. Mice with the desmin knockout gene develop normally and are fertile, however soon after birth they begin to show defects in skeletal, smooth and cardiac muscle; in particular the diaphragm and heart are affected. Mice without desmin also have impaired mitochondrial function. There are three major types of inheritance for this disease: Autosomal dominant, autosomal recessive and de novo. The most severe form is autosomal recessive and it also has the earliest onset. It usually involves all three muscle tissues and leads to cardiac and respiratory failure as well as intestinal obstruction. There is currently no cure for the disease but treatments to help the symptoms are available.

### About the Author

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