MYH9 (myosin, heavy polypeptide 9, non-muscle)

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Published in Atlas Database: August 2003
Online updated version: http://AtlasGeneticsOncology.org/Genes/MYH9ID481.html
DOI: 10.4267/2042/38010

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Identity

Other names: Myosin heavy chain, nonmuscle type A; Nonmuscle myosin heavy chain-A (NMMHC-A)
HGNC (Hugo): MYH9
Location: 22q12
Location (base pair): 33320

DNA/RNA

Description
Spans 107 kb; 40 exons

Transcription
Alternate splicing; transcripts of 4.4, 5.3 and 5.9 kb.

Protein

Description
1960 amino acids; 227 kDa (and 1752 aa, 202 kDa, and 1486 aa, 172 kDa; globular head in N-term and a coiled-coil tail in C-term; actin binding site and light chains binding site are present in the globular domain. Myosin forms hexamers with 2 heavy chains, 2 essential (alkali) light chains, and 2 regulatory light chains.

Expression
In platelets; upregulated during granulocyte differentiation (see below); also expressed in thy-mus, spleen, kidney, intestine, cochlea.

Function
Binds actin; protein of the cytoskeleton; role in cell shape and motility, and in cell division.

Mutations

Germinal
In autosomal dominant giant-platelet disorders.

Somatic
In non Hodgkin lymphomas.

Implicated in

Disease
The autosomal dominant giant-platelet disorders, May-Hegglin anomaly (MHA), Fechtner syndrome (FTNS), and Sebastian syndrome (SBS), which share a triad of thrombocytopenia, large platelets (macrothrombocytopenia (MTCP)) and characteri-stic leukocyte inclusions (Dohle-like bodies), Epstein syndrome, which associates additional Alport-like clinical features (inherited sensorineural deafness, cataracts, nephritis), and MTCP without leukocyte inclusions, as well as a nonsyndromic hereditary hearing impairment are all caused by (germinal) mutations in MYH9. These disorders appear to represent a class of allelic disorders with variable phenotypic diversity. No clear no geno-type-phenotype correlation was identified.

Anaplastic large cell lymphoma (ALCL) with t(2;22)(p23;q12) --> ALK- CLTC

Disease
ALCL are high grade non Hodgkin lymphomas; ALK+ ALCL are ALCL where ALK is involved in a fusion gene; ALK+ ALCL represent 50 to 60 % of ALCL cases (they are CD30+, ALK+); belong to the "cytoplasmic ALK+" subset.
**Prognosis**

Although presenting as a high grade tumour, a 80% five year survival is associated with this anomaly.

**Hybrid/Mutated gene**

5' MYH9 - 3' ALK

**Abnormal protein**

NH2 MYH9 - COOH ALK

**References**


Martignetti J. Five (un)easy pieces: the MYH9-related giant platelet syndromes. Haematologica. 2002 Sep;87(9):897-8


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