

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

Identity

Other names **Myosin heavy chain, nonmuscle type A**
Nonmuscle myosin heavy chain-A (NMMHC-A)

Hugo **MYH9**
 Location 22q12

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 thymus, 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 characteristic 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 genotype-phenotype correlation was identified

Entity [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 yr survival is associated with this anomaly

Hybrid/Mutated Gene
 Abnormal Protein
 5' MYH9 - 3' ALK
 NH2 MYH9 - COOH ALK

External links

Nomenclature

[Hugo](#) [MYH9](#)
[GDB](#) [MYH9](#)
[Entrez Gene](#) [MYH9_4627](#) myosin, heavy polypeptide 9, non-muscle

Cards

[Atlas](#) [MYH9ID481](#)
[GeneCards](#) [MYH9](#)
[Ensembl](#) [MYH9](#)
[CancerGene](#) [MYH9](#)
[Genatlas](#) [MYH9](#)
[GeneLynx](#) [MYH9](#)
[eGenome](#) [MYH9](#)
[euGene](#) [4627](#)

Genomic and cartography

[GoldenPath](#) [MYH9 - 22q12](#) [chr22:35001827-35108481 - 22q12.3](#) (hg17-May_2004)
[Ensembl](#) [MYH9 - 22q12.3 \[CytoView\]](#)
[NCBI](#) [Genes Cyto](#) [Gene Seq](#) [Map View - NCBI]
[OMIM](#) [Disease map \[OMIM\]](#)
[HomoloGene](#) [MYH9](#)

Gene and transcription

[Genbank](#) [Z82215](#) [SRS] [Z82215](#) [ENTREZ]
[Genbank](#) [AB191263](#) [SRS] [AB191263](#) [ENTREZ]
[Genbank](#) [AK025219](#) [SRS] [AK025219](#) [ENTREZ]
[Genbank](#) [AK025393](#) [SRS] [AK025393](#) [ENTREZ]
[Genbank](#) [AK131080](#) [SRS] [AK131080](#) [ENTREZ]
[RefSeq](#) [NM_002473](#) [SRS] [NM_002473](#) [ENTREZ]
[RefSeq](#) [NT_086921](#) [SRS] [NT_086921](#) [ENTREZ]
[AceView](#) [MYH9](#) AceView - NCBI
[TRASER](#) [MYH9](#) Traser - Stanford
[Unigene](#) [Hs.474751](#) [SRS] [Hs.474751](#) [NCBI] [HS474751](#) [spliceNest]

Protein : pattern, domain, 3D structure

[SwissProt](#) [P35579](#) [SRS] [P35579](#) [EXPASY] [P35579](#) [INTERPRO]
[Prosite](#) [PS50096 IQ](#) [SRS] [PS50096 IQ](#) [Expasy]
[Interpro](#) [IPR000048 IQ region](#) [SRS] [IPR000048 IQ region](#) [EBI]
[Interpro](#) [IPR001609 Myosin head](#) [SRS] [IPR001609 Myosin head](#) [EBI]
[Interpro](#) [IPR004009 Myosin_N](#) [SRS] [IPR004009 Myosin_N](#) [EBI]
[Interpro](#) [IPR002928 Myosin_tail](#) [SRS] [IPR002928 Myosin_tail](#) [EBI]
[Interpro](#) [IPR002017 Spectrin](#) [SRS] [IPR002017 Spectrin](#) [EBI]
[CluSTr](#) [P35579](#)
[Pfam](#) [PF00612 IQ](#) [SRS] [PF00612 IQ](#) [Sanger] [pfam00612](#) [NCBI-CDD]
[Pfam](#) [PF00063 Myosin_head](#) [SRS] [PF00063 Myosin_head](#) [Sanger]
[Pfam](#) [pfam00063](#) [NCBI-CDD]
[Pfam](#) [PF02736 Myosin_N](#) [SRS] [PF02736 Myosin_N](#) [Sanger] [pfam02736](#) [

NCBI-CDD]
[Pfam](#) [PF01576 Myosin tail 1](#) [SRS] [PF01576 Myosin tail 1](#) [Sanger]
[pfam01576](#) [NCBI-CDD]
[Prodom](#) [PD000355 Myosin head](#)[INRA-Toulouse]
[Prodom](#) [P35579 MYH9 HUMAN](#) [Domain structure] [P35579 MYH9 HUMAN](#) [sequences sharing at least 1 domain]
[Blocks](#) [P35579](#)

Polymorphism : SNP, mutations, diseases

[OMIM](#) [160775](#) [map]
[GENECLINICS](#) [160775](#)
[SNP](#) [MYH9](#) [dbSNP-NCBI]
[SNP](#) [NM_002473](#) [SNP-NCI]
[SNP](#) [MYH9](#) [GeneSNPs - Utah] [MYH9](#) [SNP - CSHL] [MYH9](#) [HGBASE - SRS]

General knowledge

[Family Browser](#) [MYH9](#) [UCSC Family Browser]
[SOURCE](#) [NM_002473](#)
[SMD](#) [Hs.474751](#)
[SAGE](#) [Hs.474751](#)
[Amigo](#) [function|ATP binding](#)
[Amigo](#) [function|actin binding](#)
[Amigo](#) [function|calmodulin binding](#)
[Amigo](#) [process|cellular morphogenesis](#)
[Amigo](#) [function|motor activity](#)
[Amigo](#) [function|motor activity](#)
[Amigo](#) [component|myosin](#)
[Amigo](#) [component|non-muscle myosin](#)
[Amigo](#) [process|perception of sound](#)
[PubGene](#) [MYH9](#)

Other databases

Probes

[Probe](#) [MYH9 Related clones \(RZPD - Berlin\)](#)

PubMed

[PubMed](#) [17 Pubmed reference\(s\) in LocusLink](#)

Bibliography

Mutations in MYH9 result in the May-Hegglin anomaly, and Fechtner and Sebastian syndromes. The May-Hegglin/Fechtner Syndrome Consortium.
Seri M, Cusano R, Gangarossa S, Caridi G, Bordo D, Lo Nigro C, Ghiggeri GM, Ravazzolo R, Savino M, Del Vecchio M, d'Apolito M, Iolascon A, Zelante LL, Savoia A, Balduini CL, Noris P, Magrini U, Belletti S, Heath KE, Babcock M, Glucksman MJ, Aliprandis E, Bizzaro N, Desnick RJ, Martignetti JA.
Nat Genet. 2000; 26: 103-105.
Medline [10973259](#)

Mutation of MYH9, encoding non-muscle myosin heavy chain A, in May-Hegglin anomaly.

Kelley MJ, Jawien W, Ortel TL, Korczak JF.
Nat Genet. 2000; 26:106-108.
Medline [10973260](#)

Human nonsyndromic hereditary deafness DFNA17 is due to a mutation in nonmuscle myosin MYH9.

Lalwani AK, Goldstein JA, Kelley MJ, Luxford W, Castelein CM, Mhatre AN.
Am J Hum Genet. 2000; 67: 1121-1128.
Medline [11023810](#)

Identification of six novel MYH9 mutations and genotype-phenotype relationships in autosomal dominant macrothrombocytopenia with leukocyte inclusions.

Kunishima S, Matsushita T, Kojima T, Amemiya N, Choi YM, Hosaka N, Inoue M, Jung Y, Mamiya S, Matsumoto K, Miyajima Y, Zhang G, Ruan C, Saito K, Song KS, Yoon HJ, Kamiya T, Saito H.
J Hum Genet. 2001; 46: 722-729.
Medline [11776386](#)

Five (un)easy pieces: the MYH9-related giant platelet syndromes.

Martignetti J.
Haematologica. 2002; 87: 897-898.
Medline [12217798](#)

MYH9-related disease: May-Hegglin anomaly, Sebastian syndrome, Fechtner syndrome, and Epstein syndrome are not distinct entities but represent a variable expression of a single illness.

Seri M, Pecci A, Di Bari F, Cusano R, Savino M, Panza E, Nigro A, Noris P, Gangarossa S, Rocca B, Gresele P, Bizzaro N, Malatesta P, Koivisto PA, Longo I, Musso R, Pecoraro C, Iolascon A, Magrini U, Rodriguez Soriano J, Renieri A, Ghiggeri GM, Ravazzolo R, Balduini CL, Savoia A.
Medicine (Baltimore). 2003; 82: 203-215.
Medline [12792306](#)

Non-muscle myosin heavy chain (MYH9): a new partner fused to ALK in anaplastic large cell lymphoma.

Lamant L, Gascoyne RD, Duplantier MM, Armstrong F, Raghav A, Chhanabhai M, Rajcan-Separovic E, Raghav J, Delsol G, Espinos E.
Genes Chromosomes Cancer. 2003; 37: 427-432.
Medline [12800156](#)

[REVIEW articles](#) *automatic search in PubMed*

[Last year publications](#) *automatic search in PubMed*

[BiblioGene - INIST](#)

Contributor(s)

Written 08-2003 Jean-Loup Huret

Citation

This paper should be referenced as such :

Huret JL . MYH9 (myosin, heavy polypeptide 9, non-muscle). Atlas Genet Cytogenet Oncol Haematol. August 2003 .

URL : <http://www.infobiogen.fr/services/chromcancer/Genes/MYH9ID481.html>

© *Atlas of Genetics and Cytogenetics in Oncology and Haematology*
