



## Anti-MLH-1 autoantibodies

### Indications

- ▶ Dermatomyositis/Polymyositis
- ▶ Aplastic Anemia

### see also

- ▶ Autoantibodies in dermatomyositis and myositis
- ▶ Autoantibodies in hematological diseases (erythrocytes)

### Antigen

PMS1 (932 amino acid; 105,8 kDa) is a member of the mutL-family of DNA mismatch and repair proteins (MMR proteins), which are responsible for recognition and repairing erroneous insertion, deletion and mis-incorporation of bases in double stranded DNA molecules (table 1). Mutation of the coding genes are associated with the elevated risk for colon carcinomas.

**Table 1** MMR proteins of *E. coli* and their human homologs

<i>E. coli</i>	Human	Remarks
mutS	MSH2 MSH3 MSH4 MSH5 MSH6	MSH: <b>mutS</b> protein <b>h</b> omolog
mutL	MLH-1 MLH-2 PMS-1 PMS-2	MLH: <b>mutL</b> -homolog  PMS: <b>postmeiotic</b> <b>s</b> egregation (proteins are also involved in crossing over during meiosis)
mutH	none	Endonuclease of <i>E. coli</i>

### Prevalence

**Table 2** Disease associations of autoantibodies against PMS1

Disease	Prevalence	Authors
Autoimmune myositis	7,5 %, *1	Casciola-Rosen et al. 2001
Aplastic anemia	10,0 % *2	Hirano et al. 2005

\*1 4 of 53 patients associated with the following autoantibodies:  
 Patient 1: M1 - 2  
 Patient 2: DNA-PK, PARP  
 Patient 3: 125 kDa protein  
 Patient 4: PMS-2, MLH1

\*2 not found in American patients.

### Methods

Immuno blot using recombinant PMS1 (Hirano et al. 2005). Radioimmunoprecipitation with <sup>35</sup>S-methionine labeled *in vitro* transcribed and translated PMS1 (Casciola-Rosen et al. 2001).

### Literature

Casciola-Rosen LA, Pluta AF, Plotz PH, Cox AE, Morris S, Wigley FM, Petri M, Gelber AC, Rosen A: The DNA mismatch repair enzyme PMS1 is a myositis-specific autoantigen. *Arthritis Rheum* (2001); 44(2): 389 - 396 (PMID: [11229471](#)).

Hirano N, Butler MO, Guinan EC, Nadler LM, Kojima S: Presence of anti-kinectin and anti-PMS1 antibodies in Japanese aplastic anaemia patients. *Br J Haematol* (2005); 128(2): 221 - 223 (PMID: [15638857](#)).