PublisherInfo		
PublisherName	:	BioMed Central
PublisherLocation	:	London
PublisherImprintName	:	BioMed Central

Hyper-IgM syndrome dissected

ArticleInfo		
ArticleID	:	4007
ArticleDOI	:	10.1186/gb-spotlight-20010309-01
ArticleCitationID	:	spotlight-20010309-01
ArticleSequenceNumber	:	78
ArticleCategory	:	Research news
ArticleFirstPage	:	1
ArticleLastPage	:	2
ArticleHistory	:	RegistrationDate : 2001–03–09 OnlineDate : 2001–03–09
ArticleCopyright	:	BioMed Central Ltd2001
ArticleGrants	:	
ArticleContext	:	130592211

Tudor Toma
Email: ttoma@mail.dntis.ro

Hyper-IgM syndrome is an immunodeficient state characterized by a normal to elevated serum concentration of IgM with low or absent IgG, IgA and IgE. A rare form of hyper-IgM syndrome, X-linked, is associated with ectodermal dysplasia (XHM-ED): the absence of hair, teeth or sweat glands. In a study in March Nature Immunology, researchers from the National Institute of Allergy and Infectious Diseases, Bethesda, identify some aspects of the mechanism that leads to the immune abnormalities seen in these patients.

Jain *et al* found that mutations in the putative zinc-finger domain of the gene encoding NEMO (nuclear factor ?B essential modulator) prevents the normal functioning of the transcription factor NF-?B. XHM-ED patients carry this mutation and as a consequence their B cells are unable to undergo immunoglobulin class-switch recombination (*Nat Immun* 2001 **2:**223-228). In addition, antigen-presenting cells were unable to synthesise the NF-?B-regulated cytokines interleukin 12 or tumour necrosis factor-a when stimulated with CD40L. These are critical cytokines in fighting bacterial infections.

Further research is required to determine what other genes are affected by this mutation in NEMO.

References

1. Jain A, Ma CA, Liu S, *et al:*Specific missense mutations in NEMO result in hyper-IgM syndrome with hypohydrotic ectodermal dysplasia. *Nat Immun* 2001 2:223-228, [http://immunol.nature.com]

2. National Institute of Allergy and Infectious Diseases, [http://www.niaid.nih.gov/default.htm]