

Hyperimmunoglobulinemia IgM syndrome/other types

In addition to the most common X-linked form caused by a mutation in the gene for **the CD40 ligand**, there are several rare forms of IgM hyperglobulinemia. The underlying clinical signs (**high IgM and** no or low levels of other Ig) are **the same** in all forms.

IgM hyperimmunoglobulinemia syndrome, type 2

(HIGM2, OMIM: 605258 (<https://omim.org/entry/605258>))

This form is caused by mutation of **AICDA** gene (**A**ctivation-**i**nduced **c**ytidine **d**eaminase, localization 12p13). Unlike the HIGM1 form, there is probably a defect in B-lymphocytes and patients are not prone to opportunistic infections. Heredity is autosomal recessive.

IgM hyperimmunoglobulinemia syndrome, type 3

(HIGM3, OMIM: 606843 (<https://omim.org/entry/606843>))

This form is caused by a mutation in the gene for **the CD40 antigen** (localization 20q12-q13.2). It is inherited as an autosomal recessive trait. The same process as in HIGM1 is disrupted, there is no isotype rearranging and similar clinical manifestations occur.

IgM hyperimmunoglobulinemia syndrome, type 4

(HIGM4, OMIM: 608184 (<https://omim.org/entry/608184>))

So far, we know the least about this form. The manifestations are similar to those of HIGM2 – but AICDA activity is maintained and the overall course is lighter (partial IgG production is preserved). It is likely to be a regulation disorder of isotype rearranging or defects in DNA recombination mechanisms.

IgM hyperimmunoglobulinemia syndrome, type 5

(HIGM5, OMIM: 608106 (<https://omim.org/entry/608106>))

This form is caused by mutation of **the UNG** gene (**U**racil-**D**N**A** glycosylase, localization 12q23-q24.1). Here too we find a defect in the isotype rearrangement and a phenotype most corresponding to the HIGM2 form.

References

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