IgM hyperimmunoglobulinemia syndrome/other types

In addition to the most common X-linked form caused by a mutation in the CD40 **ligand gene** , there are several rare forms of IgM hyperglobulinemia. The basic clinical symptoms (high level of IgM and no or low level 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 a mutation of the **AICDA gene** (Activation - induced cytidine de a minase, localization 12p13). Unlike the HIGM1 form, there is probably a defect in B-lymphocytes and patients are not prone to opportunistic infections. Inheritance 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 **CD40 antigen** gene (localization 20q12-q13.2). It is inherited in an autosomal recessive manner. Thus, the same process as for HIGM1 is disturbed, there is no isotype rearrangement and similar clinical manifestations occur.

IgM hyperimmunoglobulinemia syndrome, type 4

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

We still know the least about this form. Manifestations are similar to HIGM2 - however, AICDA activity is preserved and the overall course is milder (partial IgG production is preserved). It is likely to be disorders of isotype rearrangement regulation or defects of DNA repair mechanisms.

IgM hyperimmunoglobulinemia syndrome, type 5

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

This form is caused by a mutation of the **UNG** gene (**U**racil-D**N**A **g**lycosylase, 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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