Hyperimmunoglobulin IgM 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 IgM** and no or low levels of other Ig) are the **same in all forms**.

Hyperimmunoglobulin IgM syndrome, type 2

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

This form is caused by a mutation in the **AICDA** gene (**A**ctivation-**i**nduced **c**ytidine **d**e**a**minase, 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.

Hyperimmunoglobulin IgM 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 autosomal recessively. The same process as in HIGM1 is disrupted, there is no isotype switching and similar clinical manifestations occur.

Hyperimmunoglobulin IgM syndrome, type 4

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

We know the least about this form so far. The manifestations are similar to HIGM2 - however, the activity of AICDA is maintained and the overall course is lighter (partial IgG production is preserved). These are probably defects in the regulation of isotype switching or defects in DNA reperative mechanisms.

Hyperimmunoglobulin IgM syndrome, type 5

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

This form is caused by a mutation in 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 the phenotype most corresponding to the HIGM2 form.

Reference

- ws:Syndrom hyperimunoglobulinemie IgM
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