|  |  |  |  |
| --- | --- | --- | --- |
| Patient | 1 | 2 | 3 |
| Lymphocyte phenotpying | Normal numbers/frequencies of T/B/NK cells, normal CD4/CD8 ratio | Slightly reduced T cell numbers, preserved CD4/CD8 ratio, reduced memory B cell frequency (1% of B cells) | Normal frequencies of T/B cells, inverted CD4/CD8 ratio (0,4), reduced NK cell frequency (2%) |
| T cell proliferation | - | Normal | Normal |
| Immunoglobuline levels | Normal IgG, IgA, IgM | Normal IgG, IgA, IgM, IgE | Normal IgG, IgA, IgM, IgE |
| IgG subclasses | - | IgG4 reduced | Normal |
| Specific antibody titers | - | Normal (tetanus, diphtheria, measles, mumps, rubella) | Normal (tetanus, diphtheria, measles, mumps, rubella) |
| Autoantibodies | ANAs neg.  ANCAs neg.  Anti-GM-CSF neg. | ANAs neg.  ANCAs neg. | ANAs neg.  ANCAs neg. |
| Oxidative burst (DHR test) | - | Normal | Normal |
| Complement activity | Normal | Normal | Normal |
| Specific tests | - | SAP, XIAP expression normal  NK cell degranulation normal  Platelet granules normal (not suggestive of Hermansky Pudlak syndrome)  Hair microscopy normal (not suggestive of Chediak-Higashi or Griscelli syndrome) | - |

Table E2: Immune diagnostics in STAT3 GOF patients. ANA anti-nuclear antibodies, ANCA anti-neutrophil cytoplasmatic antibodies, CD cluster of differentiation, DHR Dihydrorhodamine, GM-CSF Granulocyte-macrophage colony-stimulating factor, Ig Immunoglobulin, SAP SLAM-associated protein, XIAP x-linked inhibitor of apoptosis