**FADD deficiency mimicking ALPS-FAS: An expanding phenotype.**

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**Table S3**: **Blood profile and immunological features of P2 at time intervals**

|  |  |  |  |
| --- | --- | --- | --- |
| **Parameter** | **8 year** | **9 year** | **Reference range** |
| **White Blood cells** | **34.11× 109/L** | 15.37 × 109/L | 10±5 × 109/L |
| **Red Blood cells** | **6.10 ×1012/L** | 5.51×1012/L | 4.6±0.6 ×1012/L |
| **Haemoglobin** | 14.30 gm/dL | 12.5 gm/dL | 13.5± 2.0gm/dL |
| **Platelets** | **649 × 109/L** | 500× 109/L | 170-450 **×** 109/L |
| **Neutrophils %** | 33% (11.1**×** 109/L) | 37% (5.7**×** 109/L) | 55-70% (2-8 × 109/L) |
| **Eosinophils %** | 0.30% (0.1 **×** 109/L) | 2.5% (0.3 **×** 109/L) | 1- 4% (0.1-1 × 109/L) |
| **Monocytes %** | 10% (3.41 × 109/L) | **8% (1.22× 109/L)** | 2-8%(0.2-1.0× 109/L) |
| **Lymphocytes%** | 57% (19.3 × 109/L) | **51.9% (7.90 ×** 109/L) | 20-40% (1-5 × 109/L) |
| **Absolute lymphocyte count(ALC)** | **19340** count/mm3 | **7977** count/mm3 | 1900-3700 count/mm3 |
| **Basic Immunological evaluation** |
| **T cell % and absolute counts (count/mm3)\*** | 66% **(12764)** | **79% (6302)** | 56-75% - (1200-2600) |
| **Th cell % and absolute counts (count/mm3)** | 30% **(5802)** | 32%**( 2553)** | 28-47% (650-1500) |
| **Tc cell % and absolute counts****(count/mm3)** | 20% **(3868)** | 28% **(2234)** | 16-30% (370-1100) |
| **B cell % and absolute counts (count/mm3)** | 32% **(6168)** | 18% **(1436)** | 14-33% - (270-860) |
| **NK cell % and absolute counts (count/mm3)** | **2%** (387) | **3%** ( 239) | 4-17% - (100- 480) |
| **Immunoglobulin levels** |
| **IgG** | 8.84 | 8.55 | 4.62-16.82 g/L |
| **IgA** | 1.18 | 0.843 | 0.34-2.74 g/L |
| **IgM** | **0.37** | **0.32** | 0.38-2.51 g/L |
| **IgE** | 17.8 | 17.8 | 0-403 IU/ml |
| **CD4 -T helper cell (Th) cell evaluation** |
| **Th naïve (CD3+CD4+CD45RA+CD27+)** | 75 | 64 | 46-99% |
| **Th central memory (CD3+CD4+CD45RA-CD27+)** | 19.71 | 18 | 0.35-100% |
| **Th effector memory (CD3+CD4+CD45RA-CD27-)** | 2.45 | 12.7 | 0.27- 18% |
| **Th TEMRA (CD3+CD4+CD45RA+CD27-)** | **2.88** | **4.2** | 0.0031-1.8% |
| **T regulatory cells (CD3+CD4+CD25high CD127low)** | **-** | 5 | 3-14% |
| **CD8- Cytotoxic T (Tc) cell evaluation** |
| **Tc naïve (CD3+CD8+CD45RA+CD27+)** | 64 | 35 | 16-100% |
| **Tc central memory (CD3+CD8+CD45RA-CD27+)** | **16.27** | **20.8** | 1-6% |
| **Tc effector memory (CD3+CD8+CD45RA-CD27-)** | 2.99 | 19.7 | 05-100% |
| **Tc TEMRA (CD3+CD8+CD45RA+CD27-)** | 16.96 | 25.8 | 15-41% |
| **B cell evaluation** |
| **B memory** **(CD19+CD27+)** | **5.05%** | **1.96%** | 7-24.3% |
| **Class switch memory (CD19+CD27+IgM- IgD-)** | 1.46 **%** | **0.98** | 1.1-6.7% |
| **CD21 low B cell** **(CD19+CD38-CD21low)** | ND | 5.4% | 0.9-7% |
| **Plasma blasts** **(CD19+ CD27+CD38 high)** | ND | 0.91% | 0.4-3.6% |
| **Transitional B cells**  | - | 0.7% | 0.6-3.4% |
| **DNT evaluation** |
| **DNTs** | **19%** | **22%** | <2.5% of total T cells |
| **B220 expression on DNTs** | **37%** | **80%** | <60% |
| **HLADR+ CD45RA+ expression on DNTs** | **22%** | **29%** | <20% |
| **Fas controlled (FC) DNTs** | **-** | **85%** | <30% |
| **Biomarker evaluation** |
| **IL-10 levels**  | **-** | 19 pg/ml | <20 pg/ml |
| **Vitamin B12 levels** | **-** | - | <1500 pg/L |
| **IL-18 levels** | - | 321pg/ml | <500 pg/ml |
| **sFASL levels** | **-** | **399 pg/ml** | <200 pg/ml |
| **sIL-2Rα** | **-** | **113509 units/ml**  | 1353-45907 units/ml |

ND: Not done