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

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**Table S4 :Blood profile and immunological features of heterozygous parents**

|  |  |  |  |
| --- | --- | --- | --- |
| **Parameter** | **Father** | **Mother** | **Reference range** |
| **White Blood cells** | 7.4 **×** 109/L | 6.8× 109/L | 10±5 × 109/L |
| **Red Blood cells** | 5.65×1012/L | 4.7×1012/L | 4.6±0.6 ×1012/L |
| **Haemoglobin** | 15.6 gm/dL | 12.3 gm/dL | 12.5±1.5gm/dL |
| **Platelets** | 115× 109/L | 309 × 109/L | 200-490 **×** 109/L |
| **Neutrophils %** | 59%**-** 4.3 **×** 109/L- | 55%- 3.7×109/L | 55-70% (1.5-8 × 109/L) |
| **Eosinophils %** | 3%-0.2 **×**109/L | 1%-0.06**×** 109/L | 1- 4% (0.1-1 × 109/L) |
| **Monocytes %** | 7%- 0.4 **×**109/L | 6%- 0.42×109/L | 2-8% (0.2-1.0) |
| **Lymphocytes%** | 31%**-** 2.2× 109/L | 38%-3× 109/L | 20-40% (6-9 × 109/L) |
| **Absolute lymphocyte count(ALC)** | 2294 count/mm3 | 2386 count/mm3 | 1400-3300 count/mm3 |
| **Basic Immunological evaluation** | | | |
| **T cell % and absolute counts (count/mm3)\*** | 72% (1652) | 81% (1933) | 49-83% - (1000-2200) |
| **Th cell % and absolute counts (count/mm3)** | 40% (918) | 49% (1169) | 27-53% (530-1300) |
| **Tc cell % and absolute counts**  **(count/mm3)** | 29%(665) | 27% (644) | 16-40% (330-920) |
| **B cell % and absolute counts (count/mm3)** | 9% (206) | 13% (310) | 2-27% - (110-570) |
| **NK cell % and absolute counts (count/mm3)** | 15% (344) | 5% (119) | 2-27% - (70-480) |
| **Immunoglobulin levels** | | | |
| **IgG** | - | - | 4-15.9 g/L |
| **IgA** | - | - | 0.17-2.90 g/L |
| **IgM** | - | - | 0.34-3.48 g/L |
| **IgE** | **-** | - | 3-423 IU/ml |
| **CD4 -T helper cell (Th) cell evaluation** | | | |
| **Th naïve (CD3+CD4+CD45RA+CD27+)** | 25 | 48 | 16-100% |
| **Th central memory (CD3+CD4+CD45RA-CD27+)** | 63 | 44 | 18-95% |
| **Th effector memory (CD3+CD4+CD45RA-CD27-)** | 12 | 8 | 1-23% |
| **Th TEMRA (CD3+CD4+CD45RA+CD27-)** | 0.1 | 0.25 | 0.0083-6.8% |
| **T regulatory cells (Tregs)**  **CD3+CD4+ CD127 low CD25 high** | 7.5% | 6.45% | 3-14% |
| **CD8- Cytotoxic T (Tc) cell evaluation** | | | |
| **Tc naïve (CD3+CD8+CD45RA+CD27+)** | 15 | 40 | 06-100% |
| **Tc central memory (CD3+CD8+CD45RA-CD27+)** | 27 | 23 | 1-20% |
| **Tc effector memory (CD3+CD8+CD45RA-CD27-)** | 16 | 13 | 14-98% |
| **Tc TEMRA (CD3+CD8+CD45RA+CD27-)** | 41 | 23 | 7-53% |
| **B cell evaluation** | | | |
| **B memory**  **(CD19+CD27+)** | **32** | **41** | 7-29% |
| **Class switch memory (CD19+CD27+IgM- IgD-)** | **16** | **27** | 0.8-9.1% |
| **CD21 low B cell**  **(CD19+CD38-CD21low)** | 2.7 | 1.7 | 0.9-7% |
| **Plasma blasts**  **(CD19+ CD27+CD38 high)** | 0.39 | **0.29** | 0.4-3.6% |
| **Transitional B cells** | 0.69 | **0.07** | 0.6-3.4% |
| **DNT evaluation** | | | |
| **DNTs** | 0.5% | 1.9% | <2.5% of total T cells |
| **B220 expression on DNTs** | 8% | 4% | <60% |
| **HLADR+ CD45RA+ DNTs** | 9.6% | 5.7% | <20% |
| **Fas controlled (FC) DNTs** | 23% | 25% | <30% |
| **Biomarker evaluation** | | | |
| **IL-10 levels** | 2 pg/ml | - | <20 pg/ml |
| **Vitamin B12 levels** | **-** | - | <1500 pg/L |
| **IL-18 levels** | 13.2 pg/ml | - | <500 pg/ml |
| **sFASL levels** | 15.8 pg/ml | - | <200 pg/ml |
| **sIL-2Rα** | 2098 | - | 1353-45907 units/ml |