

Table 1. Evaluation of the laboratory parameters of cases

Laboratory parameter	Case 1	Case 2
WBC (/mm ³)	9820 (4000- 10000)	11900 (4000- 10000)
Hb (g/dl)	9.6 (11-17.2)	11.4 (11-17.2)
PLT (/mm ³)	340000 (150000- 400000)	589000 (150000- 400000)
ANC (/mm ³)	5500 (1500-7300)	6902 (1500-7300)
ALC (/mm ³)	3900 (3200-10800)	4640 (1500-5200)
AEC (/mm ³)	196 (200-500)	238 (200-500)
ESR (mm/h)	123 (0-20)	66 (0-20)
C-RP (mg/L)	169 (0-5)	150 (0-5)
IgE (IU/ml)	17(<100)	1084-55 (<100)
IgG (mg/dl)	1020 (280-666)	1830 (613-1030)
IgM (mg/dl)	252 (19-94)	146 (58-126)
IgA (mg/dl)	28 (5-39)	55 (26-80)
AntiHBs (mIU/mL)	223 (>10)	100 (>10)
Anti-B (isohemagglutinin titer)	Not tested	Anti-A: 1/64, Anti-B: 1/4 (<1/10)
Mumps IgG	Not tested	5 (0-1.1)
Varicella virus IgG (IU/ml)	Not tested	23.6 (<9)
Procalcitonin (ng/mL)	37.2 (<0.5)	Not tested
CD3+T (%)	60 (51-79)	74 (58-82)
CD3+CD4+T (%)	42 (31-54)	42 (26-48)
CD3+CD8+T (%)	18 (10-31)	28 (16-32)
CD45RA (%)	86 (72-93)	57(66-95)
CD45RO (%)	3 (9-31)	30 (14-44)
CD19+B (%)	29 (14-44)	1-14 (10-30)
CD16+56 (%)	6 (5-23)	4
CD25 activation	Normal	Normal
Neutrophil functions	Normal (DHR)	Normal (NBT)
IL-6 response after stimulation with LPS or IL-1b	defective	defective
Microorganisms detected	<i>Rotavirus</i> (stool), <i>P. aeruginosa</i> (CSF) Yeast (urine)	<i>C. albicans</i> (throat), <i>Salmonella CI</i> (stool and blood), <i>Enterococcus faecalis</i> (urine), <i>Staph. Hominis</i> (blood)
Mutation	Missense mutation in exon 3 of <i>IRAK4 gene</i> (c.161G>A, p.R54K, at protein level)	Missense mutation in exon 3 of <i>IRAK4 gene</i> (c.161G>A, p.R54K, at protein level)

WBC: White blood cell, Hb: hemoglobin, PLT: Platelet, ANC: Absolute neutrophil count, ALC: Absolute lymphocyte count, AEC: Absolute eosinophil count, ESR: Erythrocyte sedimentation rate, C-RP: C reactive protein, CD25 activation: CD25 activation of T cells after stimulation with PHA, Neutrophil functions: Chemotaxis, phagocytosis and burst functions of neutrophils, CD62L: CD62 ligand, DHR: Dihydrorhodamine (DHR) Flow Cytometric Assay, NBT: Nitroblue tetrazolium test