

**Table 2: Summary of the functional studies in the patients' cells**

Patient (Kindred)	Mutation	PKC $\delta$ expression	p40 <sup>phox</sup> phosphorylation	ROS production in circulating phagocytes	ROS production in monocyte-derived cells
<b>P1 (A)</b>	p.R614W/p.R614W	Impaired (EBV-B cells)	Impaired after PMA and Pansorbin (EBV-B cells)	NT	Impaired after PMA, PMA + IFN- $\gamma$ and PMA+LPS (MDMs and MDDCs)
<b>P2 (B)</b>	p.G248S/p.G248S	Normal (EBV-B cells and MDMs)	Impaired after PMA and Pansorbin (EBV-B cells and MDMs)	Impaired after PMA (neutrophils and monocytes)	Impaired after PMA, PMA + IFN- $\gamma$ and PMA+LPS (MDMs and MDDCs)
<b>P3 (C)</b>	p.G510S/p.G510S	Impaired (EBV-B cells, neutrophils and MDMs)	Impaired after PMA and Pansorbin (EBV-B cells and MDMs)	Impaired after PMA and <i>E. coli</i> (neutrophils and monocytes)	Impaired after PMA, PMA + IFN- $\gamma$ and PMA+LPS (MDMs and MDDCs)
<b>P4 (C)</b>	p.G510S/p.G510S	NT	NT	NT	NT
<b>P5 (C)</b>	p.G510S/p.G510S	Impaired (EBV-B cells)	Impaired after PMA and Pansorbin (EBV-B cells)	Impaired after PMA (neutrophils and monocytes)	Impaired after PMA, PMA + IFN- $\gamma$ and PMA+LPS (MDMs and MDDCs)
<b>P6 (D)</b>	c.1352+1G>A/c.1352+1G>A	Abolished (EBV-B cells)	Impaired (EBV-B cells)	Impaired after PMA (neutrophils and monocytes)	Impaired after PMA, PMA + IFN- $\gamma$ and PMA+LPS (MDMs and MDDCs)
<b>P7 (E)</b>	p.G432W	NT	NT	NT	NT
<b>P8 (E)</b>	p.G432W	NT	NT	NT	NT
<b>P9 (E)</b>	p.G432W	NT	NT	NT	NT
<b>P10 (F)</b>	c.788-2A>G/p.Q191*	Abolished (EBV-B cells)	Impaired after PMA and Pansorbin (EBV-B cells)	Impaired after PMA (neutrophils and monocytes)	Impaired after PMA, PMA + IFN- $\gamma$ and PMA+LPS (MDMs and MDDCs)
<b>P11 (F)</b>	c.788-2A>G/p.Q191*	Abolished (EBV-B cells)	Impaired after PMA and Pansorbin (EBV-B cells)	Impaired after PMA (neutrophils and monocytes)	Impaired after PMA, PMA + IFN- $\gamma$ and PMA+LPS (MDMs and MDDCs)
<b>P12 (G)</b>	c.1293_1294insA/c.1293_1294insA	NT	NT	NT	NT
<b>P13 (H)</b>	c.571+2dup/c.571+2dup	Abolished (EBV-B cells, neutrophils, monocytes and MDMs)	Impaired after PMA and Pansorbin (EBV-B cells)	Impaired after PMA (neutrophils and monocytes)	Impaired after PMA, PMA + IFN- $\gamma$ and PMA+LPS (MDMs and MDDCs)
<b>P14 (I)</b>	c.1384C>T/ c.1384C>T	Abolished (MDMs)	Impaired (MDMs)	Impaired after PMA and <i>E. coli</i> (neutrophils and monocytes)	Impaired after PMA and PMA + IFN- $\gamma$ (MDMs)
<b>P15 (I)</b>	c.1384C>T/ c.1384C>T	NT	NT	Impaired after PMA and <i>E. coli</i> (neutrophils and monocytes)	Impaired after PMA and PMA + IFN- $\gamma$ (MDMs)
<b>P16 (J)</b>	c.642del/c.642del	Abolished (MDMs)	Impaired (MDMs)	Impaired after PMA and <i>E. coli</i> (neutrophils and monocytes)	Impaired after PMA and PMA + IFN- $\gamma$ (MDMs)
<b>P17 (J)</b>	c.642del/c.642del	Abolished (MDMs)	Impaired (MDMs)	Impaired after PMA and <i>E. coli</i> (neutrophils and monocytes)	Impaired after PMA and PMA + IFN- $\gamma$ (MDMs)

NT: not-tested, MDMs: monocyte-derived macrophages, MDDCs: monocyte-derived dendritic cells, PMA: phorbol 12-myristate 13-acetate, EBV-B cells: Epstein-Barr virus-immortalized B cells, LPS: lipopolysaccharides

This table summarizes functional results obtained in cell lines (EBV-B cells) or primary cells (neutrophils, monocytes, MDMs and MDDCs).