***Supplementary Material***

**Family history of early infant death correlates with earlier age at diagnosis but not shorter time to diagnosis for severe combined immunodeficiency.**

**Anderson Dik Wai Luk1, Pamela P. Lee1, Huawei Mao1,2, Koon-Wing Chan1, Xiang Yuan Chen3, Tong-Xin Chen4, Jian Xin He5, Nadia Kechout6, Deepti Suri7, Yin Bo Tao3, Yong Bin Xu8, Li Ping Jiang9, Woei Kang Liew10, Orathai Jirapongsananuruk11, Tassalapa Daengsuwan12, Anju Gupta7, Surjit Singh7, Amit Rawat7, Amir Hamzah Abdul Latiff13, Anselm Chi Wai Lee14, Lynette P Shek15, Thi Van Anh Nguyen16, Tek Jee Chin17, Yin Hsiu Chien18, Zarina Abdul Latiff19, Thi Minh Huong Le16, Nguyen Ngoc Quynh Le16, Bee Wah Lee15, Qiang Li20, Dinesh Raj21, Mohamed-Ridha Barbouche22, Meow-Keong Thong23, Maria Carmen D. Ang24, Xiao Chuan Wang25, Chen Guang Xu26, Hai Guo Yu27, Hsin-Hui Yu18, Tsz Leung Lee1, Felix Yat Sun Yau28, Wilfred Hing-sang Wong1, Wenwei Tu1,2, Wangling Yang1,2, Patrick Chun Yin Chong1, Marco Hok Kung Ho1, Yu Lung Lau1,2\***

**\*Correspondence:** Yu Lung Lau, MD (Honors), Department of Paediatrics & Adolescent Medicine, Li Ka Shing Faculty of Medicine, the University of Hong Kong, Pokfulam Road, Hong Kong Special Administrative Region, PR China:[lauylung@hku.hk](mailto:lauylung@hku.hk)

**Supplementary table E4. Characteristics of patients with genotype-immunophenotype miscorrelation.**

Patient Clinical presentation ALC(x109/L) FH CD19+cell/uL(%) Mutation(Gene) Other SCID gene tested negative

P001 Lymphopenia 0.14 No 70(50) c.3G>T; p.M1I *(IL2RG)* Not done

P002 Lymphopenia, pneumonia, CMV infection 0.67 No 13.4(2) c.127delA; p.T43fsX70 *(IL2RG)* Not done

P003 Chronic diarrhea, FTT, disseminated BCG 3.6 Yes2 0(0) c.202G>T; p.E68X *(IL2RG)* Not done

P005 Recurrent infections 0.18 No 3.6(2) c.202G>A; p.E68K *(IL2RG)* *DCLRE1C, RAG1, RAG2*

P0781 Severe eczema, RTI, recurrent salmonella GE, 7.64 No 267.4(3.5) c.1178delG; p.G393fsX402 *(RAG1)* Not done

recurrent herpes zoster, eosinophilia c.2095C>T; p.R699W *(RAG1)*

B- SCID was defined as having <134 CD19+ cells/uL. c. indicates nucleotide changes; p. indicates predicted changes in protein. ALC, absolute lymphocyte count; FH, family history of early infant death; CMV, cytomegalovirus; FTT, failure to thrive; RTI, respiratory tract infection; GE, gastroenteritis. Lymphopenia was defined as ALC below 3 x 109/L. 1P078 had double heterozygous mutation of *RAG1*. 2P003 had family history of early infant death of 5 maternal uncles.