

Appendix S1

## MATERIAL AND METHODS

### *Ethical considerations*

The patient, now 26 years old, was first investigated at the Queen Silvia Children's Hospital, Gothenburg at the age of 3 and hence, from the age of 17, investigations and treatments were carried out at Uppsala University Hospital. All analyses were performed according to the Declaration of Helsinki and were part of our efforts to correctly diagnose and treat the patient to the best of our knowledge; informed consent was obtained from both parents and later also from the patient.

### *Granulocyte cell surface expression*

Leukocytes from 1 ml of heparinised blood were prepared, labelled and analysed by flow cytometry according to procedures previously described (8). FITC-labelled monoclonal antibodies against CD11a clone MHM24 (Dakocytomation (DC)), CD11b clone Bear 1 (Beckman Coulter (BC)), CD11b clone 2LPM19c (DC), CD11c clone KB90 (DC), CD18 clone MHM23 (DC), CD18 clone 7E4 (BC), CD18 clone 6.7 (BD Biosciences), CD16 clone DJ130c (DC), CD32 clone FLI8.26 (BD), CD64 clone 22 (BC), CD65 clone 88H7 (BC) and CD162 clone 5D8-8-12 (BC). Flow cytometric analysis was carried out on an EPICS XL flow cytometer (BC). Cell surface expression is presented as the mean fluorescence intensity of the granulocyte population and the relative number of positive granulocytes (%) i.e. granulocytes with an expression exceeding that of the isotype control.

### *Granulocyte migration*

Granulocyte migration assays were performed in a micro Boyden chamber by a modification of the Boyden chamber method (9). Granulocytes were isolated from heparinised blood by means of dextran sedimentation. Micropore filters approximately 150 µm thick with a pore size of 3 µm were used (Millipore Corp., Bedford, MA). C5a ( $10^{-8}$  mol/l) and interleukin-8 (IL-8) ( $10^{-9}$  mol/l) were used as chemotactic stimuli. Incubation was performed for 1 h and after fixation and staining migration of granulocytes was assayed by means of the leading front technique (10).

### *Granulocyte respiratory burst*

Analysis of granulocyte respiratory burst induced by *E. coli* and PMA, respectively, was performed by use of the Phagoburst test (Glycotope Biotechnology, Heidelberg, Germany) with the method described by the manufacturer.

### *Granulocyte phagocytosis*

Phagocytosis of *E. coli* was measured by the use of FITC-labelled bacteria (Pinnacle Biosystems Inc, Burlingame, CA, USA) by the procedure suggested by the manufacturer.

### *Mutation analysis*

Sequence analysis of *ITGB2* was performed according to Roos et al. (11). Primer sequences of *PSTPIP1* are available upon request.