

Supplemental Data

Bi-allelic Variants in the GPI Transamidase Subunit

PIGK Cause a Neurodevelopmental Syndrome with

Hypotonia, Cerebellar Atrophy, and Epilepsy

Thi Tuyet Mai Nguyen, Yoshiko Murakami, Sabrina Mobilio, Marcello Niceta, Giuseppe Zampino, Christophe Philippe, Sébastien Moutton, Maha S. Zaki, Kiely N. James, Damir Musaev, Weiyi Mu, Kristin Baranano, Jessica R. Nance, Jill A. Rosenfeld, Nancy Braverman, Andrea Ciolfi, Francisca Millan, Richard E. Person, Ange-Line Bruel, Christel Thauvin-Robinet, Athina Ververi, Catherine DeVile, Alison Male, Stephanie Efthymiou, Reza Maroofian, Henry Houlden, Shazia Maqbool, Fatima Rahman, Nissan V. Baratang, Justine Rousseau, Anik St-Denis, Matthew J. Elrick, Irina Anselm, Lance H. Rodan, Marco Tartaglia, Joseph Gleeson, Taroh Kinoshita, and Philippe M. Campeau

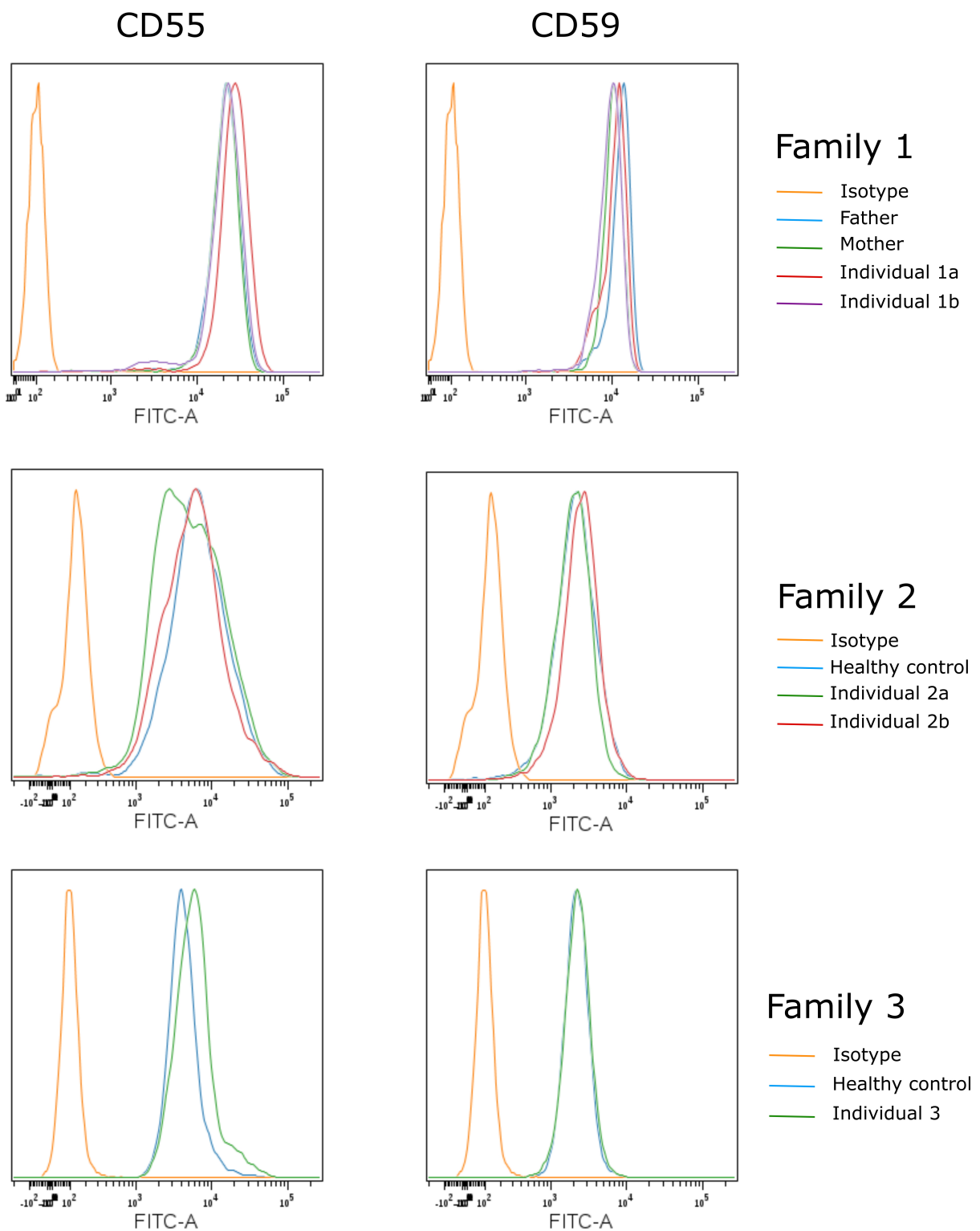


Figure S1: Cell surface levels of other GPI-APs in blood cells of affected individuals. Blood samples of the individuals in families 1, 2 and 3 and control cells were stained with CD55 and CD59. Figure shows representative analysis of cell surface GPI-AP levels of granulocytes from triplicate experiments.

Figure S1