

Supplemental Data

Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders

Erfan Aref-Eshghi, Jennifer Kerkhof, Victor P. Pedro, Groupe DI France, Mouna Barat-Houari, Nathalie Ruiz-Pallares, Jean-Christophe Andrau, Didier Lacombe, Julien Van-Gils, Patricia Fergelot, Christèle Dubourg, Valérie Cormier-Daire, Sophie Rondeau, François Lecoquierre, Pascale Saugier-Veber, Gaël Nicolas, Gaetan Lesca, Nicolas Chatron, Damien Sanlaville, Antonio Vitobello, Laurence Faivre, Christel Thauvin-Robinet, Frédéric Laumonnier, Martine Raynaud, Mariëlle Alders, Marcel Mannens, Peter Henneman, Raoul C. Hennekam, Guillaume Velasco, Claire Francastel, Damien Ulveling, Andrea Ciolfi, Simone Pizzi, Marco Tartaglia, Solveig Heide, Delphine Héron, Cyril Mignot, Boris Keren, Sandra Whalen, Alexandra Afenjar, Thierry Bienvenu, Philippe M. Campeau, Justine Rousseau, Michael A. Levy, Lauren Brick, Mariya Kozenko, Tugce B. Balci, Victoria Mok Siu, Alan Stuart, Mike Kadour, Jennifer Masters, Kyoko Takano, Tjitske Kleefstra, Nicole de Leeuw, Michael Field, Marie Shaw, Jozef Gecz, Peter J. Ainsworth, Hanxin Lin, David I. Rodenhiser, Michael J. Friez, Matt Tedder, Jennifer A. Lee, Barbara R. DuPont, Roger E. Stevenson, Steven A. Skinner, Charles E. Schwartz, David Genevieve, and Bekim Sadikovic

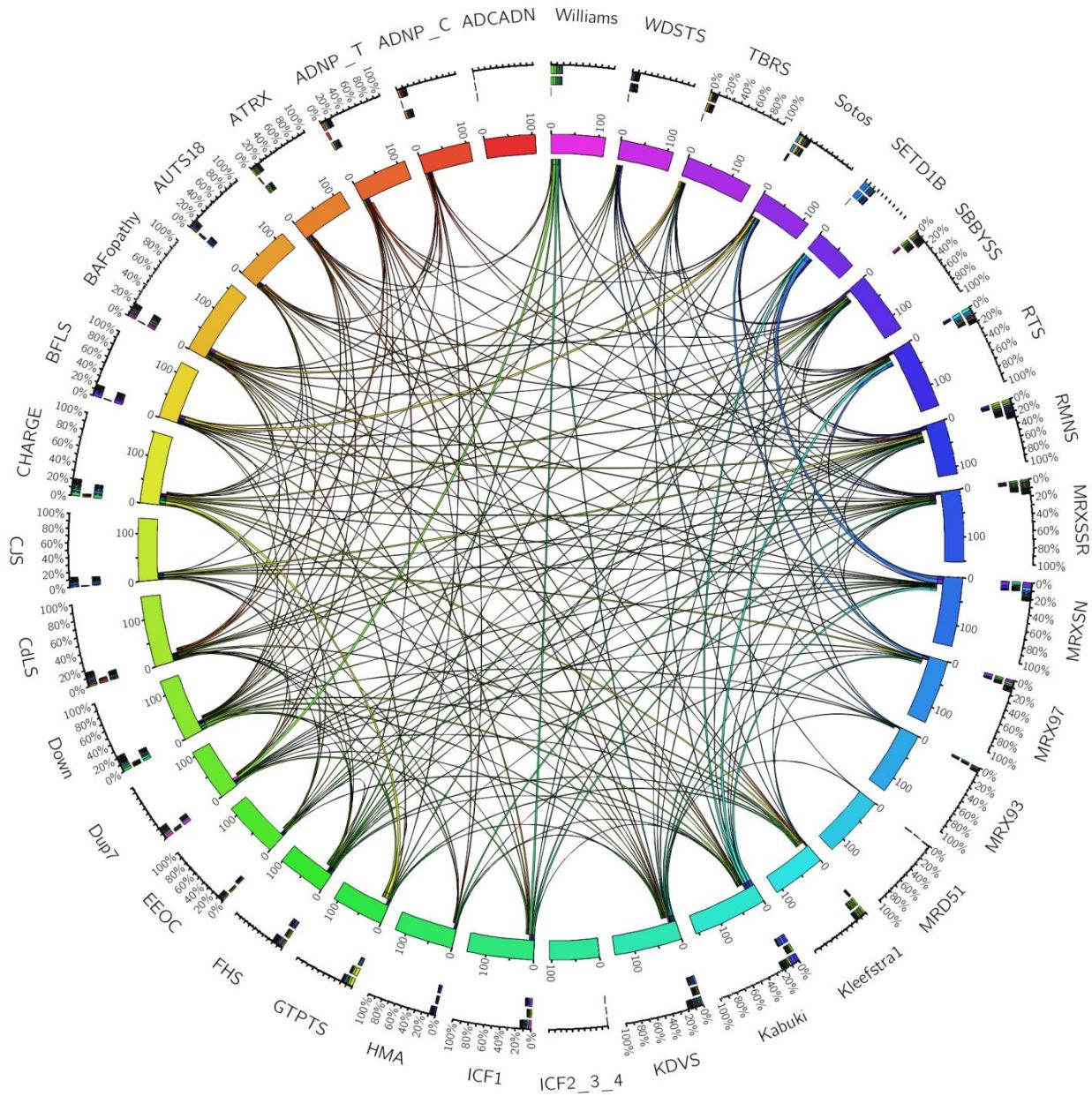


Figure S1- Co-occurrence of CpG sites between different episignatures

Circos plot showing the number of probes co-occurring between any two episignatures as demonstrated using the thickness of the connecting lines; The probe counts per episignature is scaled from 0 to 100%. The connecting lines show the percentage of probes from each signatures being shared with the others. All of the shared components are summed as stacks of bars on the right side of each episignature. As seen, only a small fraction of differentially methylated probes are common among disorders and the portion of shared probes from each episignatures with all others does not reach 5%.