Dr. William T. Gibson, MD, PhD(Cantab), FRCPC, FCCMG, is a Professor of Medical Genetics at the University of British Columbia, and a Senior Clinician Scientist at British Columbia Children's Hospital Research Institute in Vancouver, Canada. He is also an overseas member of the Royal Society of Medicine. His team recruits patients and families with rare genetic diseases, in order to solve the underlying genetic cause by finding different mutations in the same gene among unrelated families with similar diseases.

The Gibson lab has been successful in finding mutations in major “epigenes” – these are genes that code for proteins that open DNA up to activate it, or close it down to keep it quiet. We hope that the rare syndromes we have described will help us learn how these epigenetic programs can increase or decrease the risks for common diseases in the general population.

Our group’s work has already been put into practice by key knowledge users. Labs that offer clinical exome sequencing and/or multi-gene panels now report out mutations in EZH2, EED, SUZ12 and SETD1B as disease-causing. Impact on families includes better diagnosis, with better access to health care, education, and accurate genetic counselling.