A CLINICAL METHOD FOR GENOTYPING LARGE GENES FOR MUTATIONS THAT POTENTIALLY CAUSE DISEASE

A method of determining polymorphisms within a large gene comprising the steps of: (a) making a Whole-Genome Amplification (WGA) to obtain sufficient amounts of genetic templates for DNA analysis; (b) enriching the WGA sample with nested primers designed for the large gene; (c) using the enriched WGA sample for high resolution melt (HRM); and (d) detecting differential melt profiles during the transition from double strand to single strand with an increase in temperature wherein sequence point mutations within the gene affects the thermal stability and gives a different melt profile from the normal non-mutated gene sequence, and kits to carry out detection of the same

Figure 1