

Urine GC/MS, dried blood spots MS/MS and serum total homocysteine test

MMA combined with HC

Isolated MMA

PCR-HRM analysis
for MMACHC

Next-generation sequencing

Compound
heterozygous mutations

Negative

Single
heterozygous mutation

Sanger sequencing validation

Specially designed method for
mutation c.609G>A (p.Trp203Ter)

Next-generation sequencing

Homozygous difference plots

Indifference plots

Sanger sequencing validation

Next-generation sequencing

