

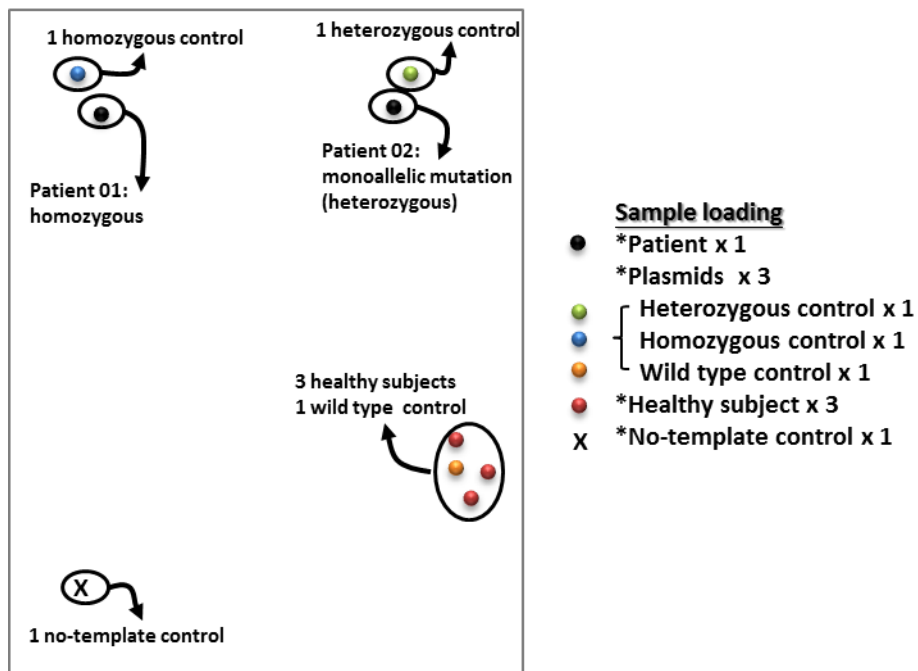
Supplementary Information

Table of content

Supplementary Figure 1. Example and interpretation of allelic discrimination using the hotspot-based TaqMan assay in newly-diagnosed GS patients.

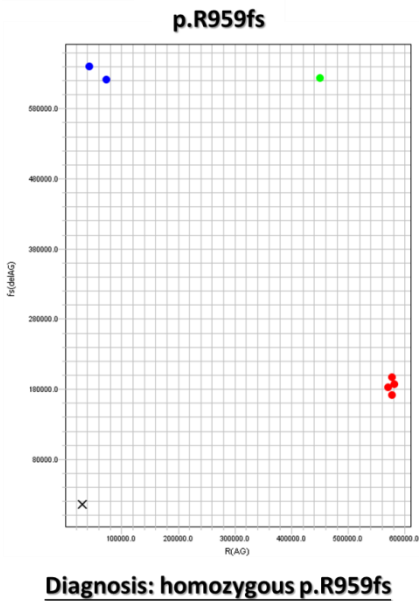
Supplementary Figure 2. Interpretation of validation in 12 newly-diagnosed GS patients.

Supplementary Figure 1. Example and interpretation of allelic discrimination using the hotspot-based TaqMan assay in newly-diagnosed GS patients

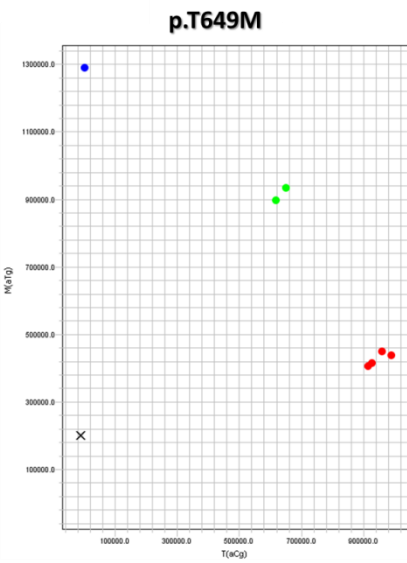
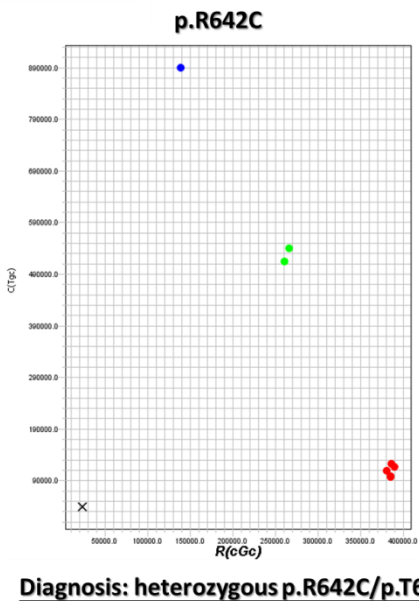


Supplementary Figure 2. Interpretation of validation in 12 newly-diagnosed GS patients.

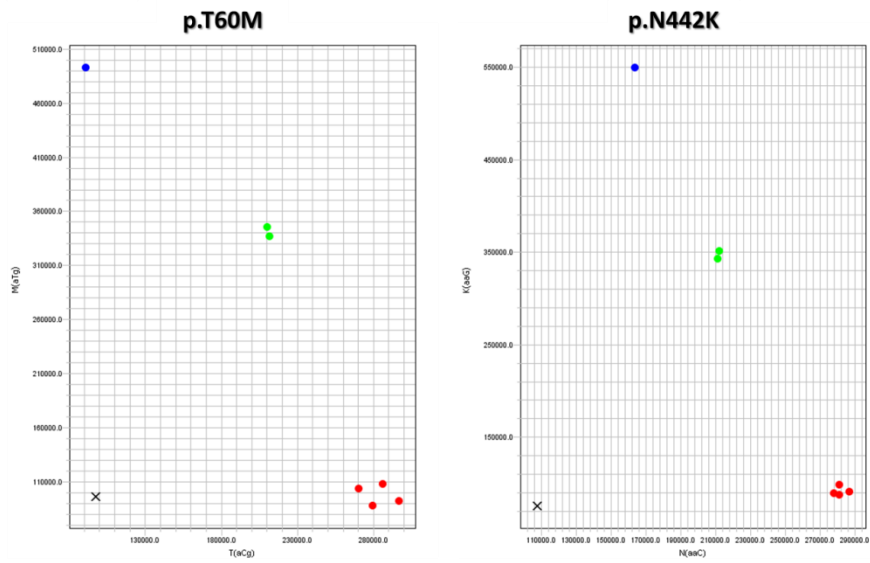
Case 1



Case 2

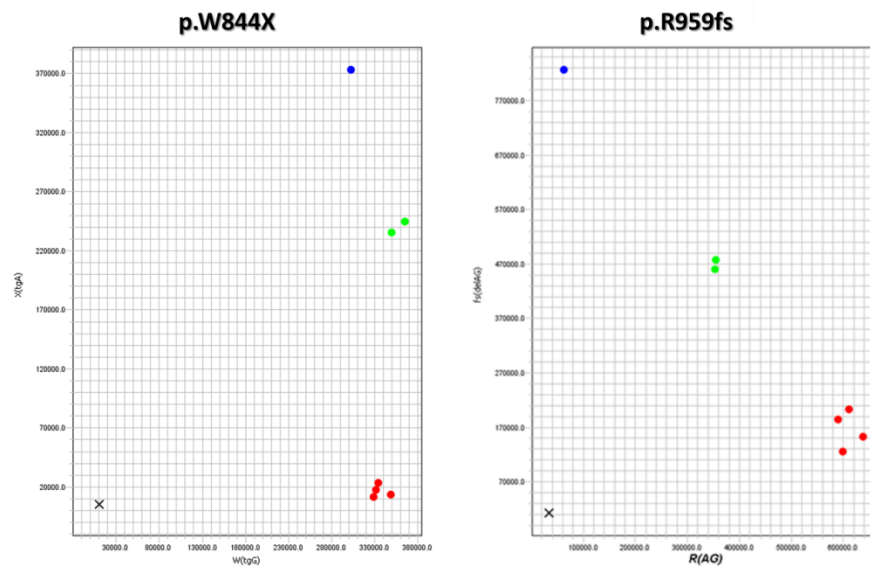


Case 3



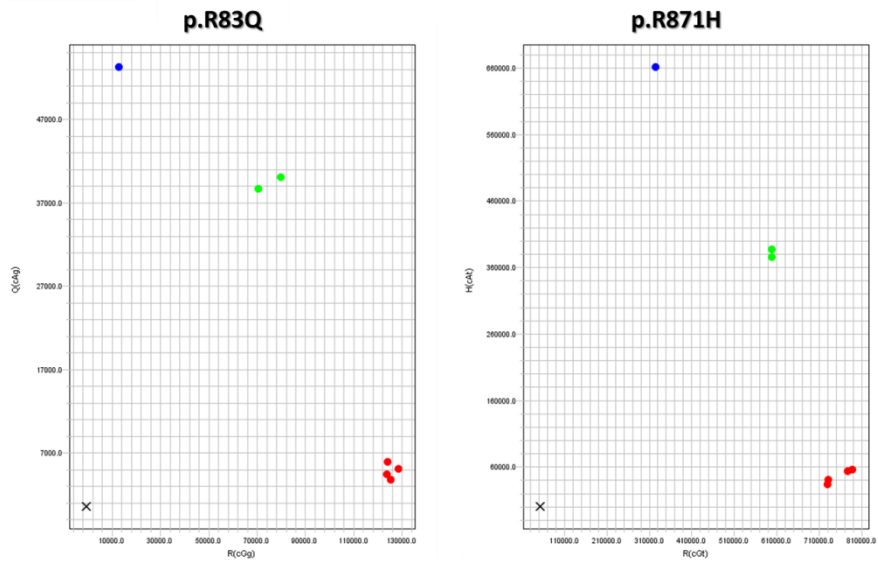
Diagnosis: heterozygous p.T60M/p.N442K

Case 4



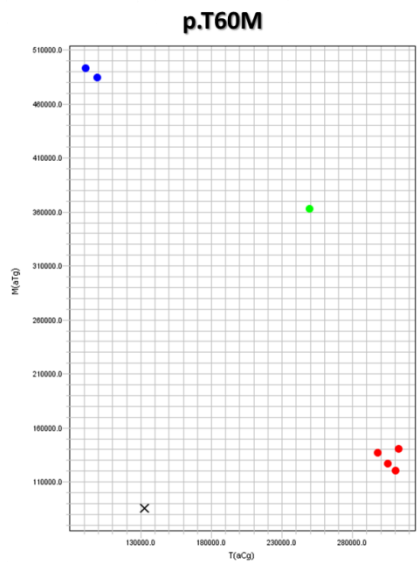
Diagnosis: heterozygous p.W844X/p.R959fs

Case 5



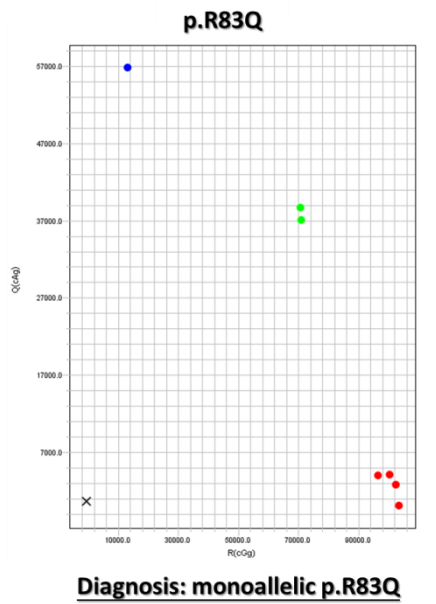
Diagnosis: heterozygous p.R83Q/p.R971H

Case 6

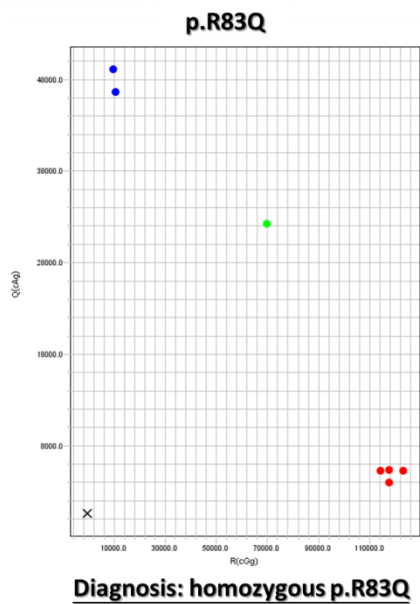


Diagnosis: homozygous p.T60M

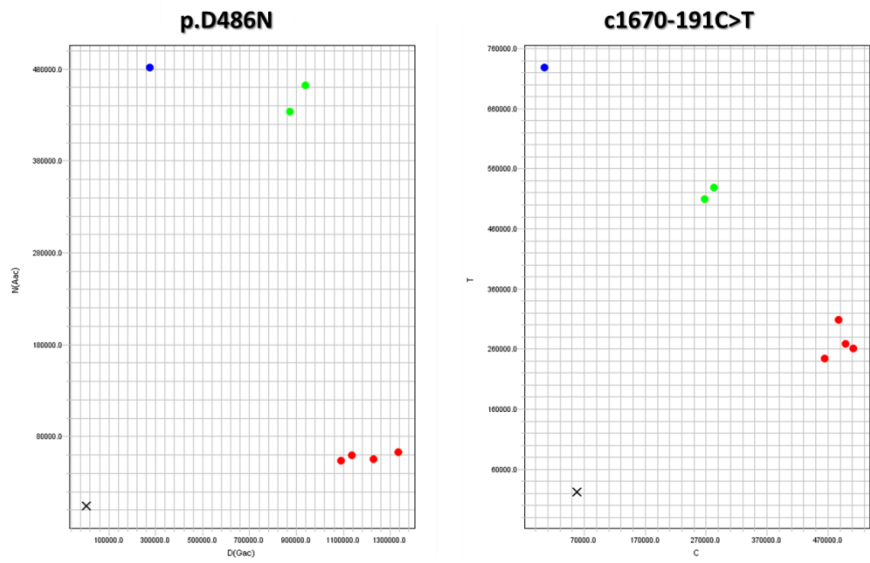
Case 7



Case 8

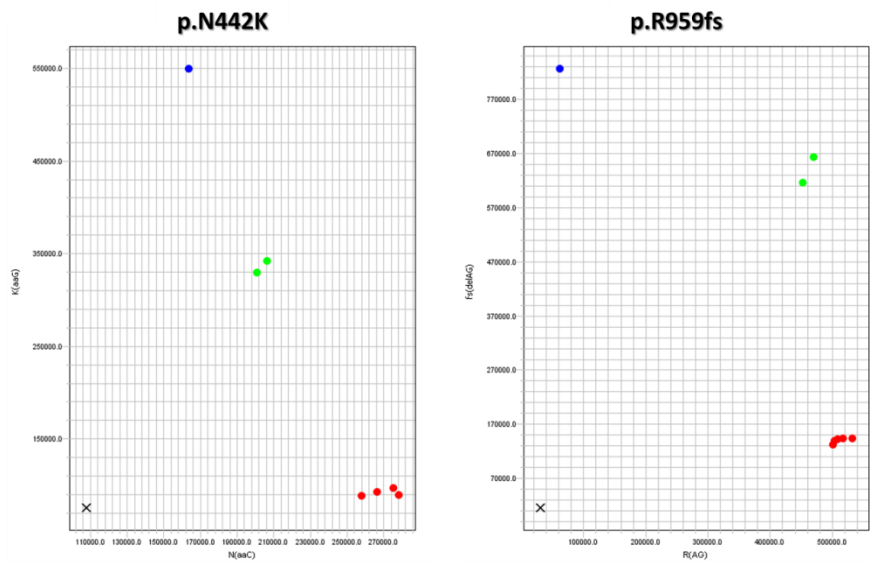


Case 9



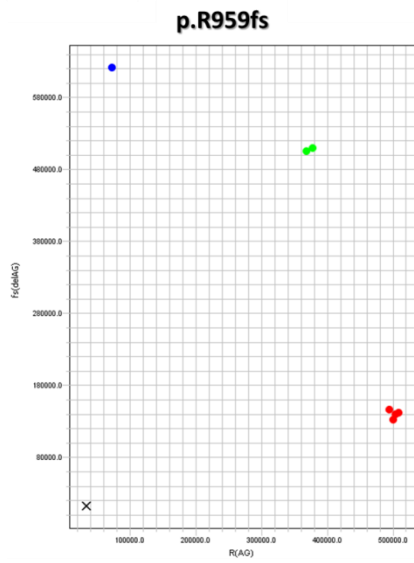
Diagnosis: heterozygous p.D486N/c1670-191C>T

Case 10



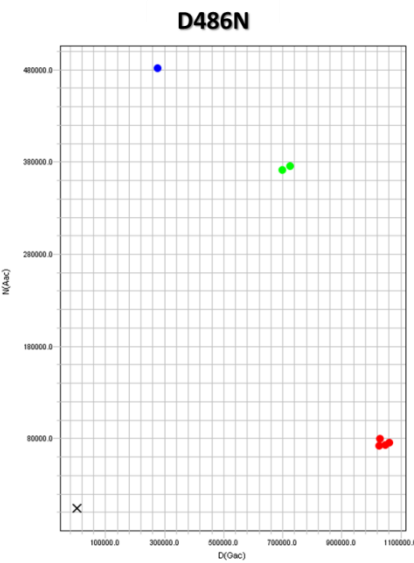
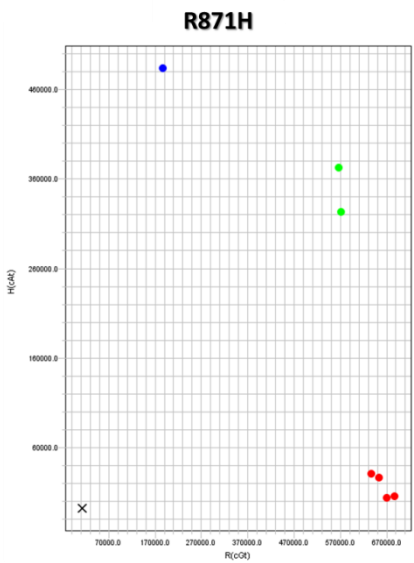
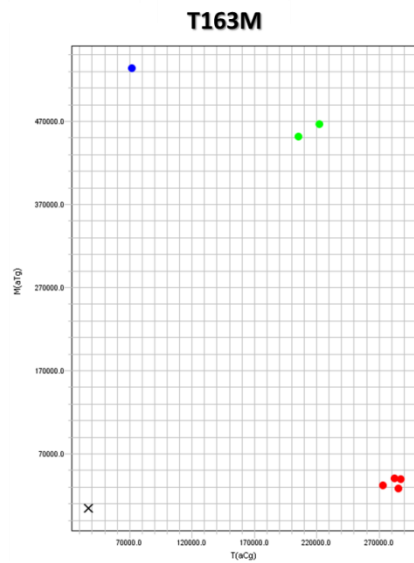
Diagnosis: heterozygous p.N442K/p.R959fs

Case 11



Diagnosis: monoallelic p.R959fs

Case 12



Diagnosis: triple p.T163M/p.R871H/p.D486N