Based on report number:

Relevant gene(s):

Patient diplotype:

Patient phenotype:

Case 1: Cystic Fibrosis

VS is a 6-year-old Caucasian female who was recently diagnosis with Cystic Fibrosis after an elevated sweat chloride test along with a confirmative genetic screening. VS’s parent (neither of which have the disease) are very worried about their daughter, as the little information they know about the disease state and VS’s mortality is very negative. They are currently expecting another child, and would like to have a large family, but are worried about potentially having more children with the condition.

You are consulted by a local pulmonologist to help determine if a treatment regimen containing ivacaftor would be appropriate for VS, as well give the family some education upon their daughters new diagnosis, and how it may affect their family.

Recommendation:

Note:

Recommendation based on:

Found at: