MVP-ROAR-FH Sample Post-counseling CPRS Note

The following result note was placed in the <FACILITY NAME> CPRS medical record as part of a remote encounter and can be viewed via VistaWeb/Joint Legacy Viewer. A copy was also placed in the patient-side CPRS for the convenience of the provider and the Veteran. Any requested addendum to this note must also be communicated to <FACILITY NAME> via email so the original note is modified as appropriate.

GENETIC TEST RESULT NOTE
[X] Via Telephone

RESULTS: Genetic analysis (next generation sequencing and deletion/duplication analysis) for mutations in four genes associated with hypercholesterolemia was POSITIVE for a heterozygous pathogenic variant in the LDLR gene (c.661G>T; p.Asp221Tyr). This result explains the Veteran's history of early onset hypercholesterolemia with elevated direct LDL levels.

The Veteran was contacted with this information on XXXX and had the opportunity to ask questions.

A copy of these results can be found in the <LOCAL FACILITY> CPRS or Vista Imaging; please contact the <LOCAL FACILITY> if results cannot be found.

INTERPRETATION/DISCUSSION: Familial hypercholesterolemia (FH) is a common hereditary condition with an estimated prevalence of about 1/300. Mutations in the LDLR gene are associated with autosomal dominant FH. Penetrance of the condition is estimated to be >90%. Those with an FH mutation are at an increased risk of developing high cholesterol, particularly very elevated LDL-C levels. This can lead to plaque deposition increasing the risk for cardiovascular disease -especially coronary artery disease (CAD)- and death. Untreated men are at a 50% risk of having a coronary event by age 50. Some individuals have secondary physical findings such as xanthomas and corneal arcus.

60-80% of FH cases are attributed to mutations in the LDLR gene. Most cases of FH are inherited; it is likely one of the Veteran's parents carries the same mutation. Therefore, there is a 50% chance his full siblings have the familial LDLR mutation. There is also a 50% chance Mr. X's future children could have the same LDLR mutation.

Those with homozygous FH are more severely affected and are at higher risk for cardiac events at a young age. Therefore, the Veteran may want to consider discussing preconception carrier testing with his partner to determine if they are at an increased risk of having a child affected with homozygous FH.

MEDICAL MANAGEMENT:

Per a XXXX neurology note, the Veteran reports he is taking Atorvasatin and reports he has restarted smoking 1ppd. On XXXX his cholesterol serum levels were reported to be 315mg/dl and his direct LDL was reported to 133 mg/dl.

Literature and professional organization guidelines recommend the following in general for individuals with FH. His personal health care team should modify to meet this Veteran's specific needs.
- Lifestyle modification should focus on regular physical activity and weight control. A healthy diet with reduced saturated fat intake and increased (10-20 g/day) intake of soluble fiber is recommended. Limit alcohol consumption. Affected individuals should NOT use tobacco products.
- If he is not currently taking them, consider use of statins to reduce LDL-C levels if levels are >100mg/dL.
- If at a high risk for CAD or stroke, use of low dose aspirin (75-81 mg/day) can be considered.
- Blood pressure should be monitored and treated if >140/90mm Hg.
- Diabetes should be screened for and managed appropriately, if diagnosed.
- If statins are not working, consideration of other drug therapies and/or LDL apheresis should be considered. These may include cholesterol absorption inhibitors, Mipomersen (to decrease APOB production), MTP inhibitors, PCSK9 inhibitors, bile acid sequestrants and stanol esters (to decrease cholesterol absorption and increase LDLR activity).

PLAN:
- Local providers will continue to monitor the Veteran's LDL-C levels and monitor for any cardiac complications.
- Primary care: please discuss recommended lifestyle modifications with the Veteran and review the efficacy of his statin therapy.
- Primary care: consider referral to a lipid specialist if LDL-C concentrations cannot be reduced after treatment with statins and lifestyle modifications.
- No further germline genetic testing is recommended for the Veteran at this time.
- The Veteran will be mailed a copy of his results and a letter summarizing them.
- Mr. X was encouraged to share this information with his family members as his first-degree relatives are all genetic testing candidates. Testing of second-degree relatives is also recommended, however it is unclear at this time what side of the family the Veteran inherited the mutation from. Invitae offers free testing to close relatives of those with an identified pathogenic mutation if tested prior to XXXX. Blood relatives are encouraged to have regular cholesterol level screening.

Duration of Call: 7 minutes