



## The Apo E Genotype Test for Type III Hyperlipoproteinemia

Type III hyperlipoproteinemia (broad beta disease) should be considered in all patients with elevation of both serum cholesterol and triglycerides. The DNA test for this lipid disorder accurately determines the apolipoprotein E (apo E) genotype. The apo E genotype E2/E2 is diagnostic of type III hyperlipoproteinemia in individuals who have elevated serum cholesterol and triglycerides and accumulation of the abnormal lipoprotein,  $\beta$ -VLDL. Apo E genotyping is also of value for risk determination in relatives. Type III hyperlipoproteinemia is associated with accelerated atherosclerosis, coronary heart disease and peripheral vascular disease.

### Indications for Apo E Genotype Testing:

- Total serum cholesterol >240 mg/dl and triglycerides (TG) >150 mg/dl  
**OR**  
LDL cholesterol >[age + 100] and TG >[age + 100]  
**PLUS**  
VLDL cholesterol/TG ratio >0.3,  
**OR**  
 $\beta$ -VLDL revealed by lipoprotein electrophoresis as a "broad beta band"

Patients may present with:

- No symptoms
- Peripheral vascular disease
- Coronary artery disease (coronary heart disease)
- Cutaneous xanthomas
- Diabetes mellitus, obesity or hypothyroidism (conditions which can trigger type III hyperlipoproteinemia in individuals with the apo E genotype E2/E2)

- Relatives of individuals diagnosed with type III hyperlipoproteinemia, especially siblings

### Recommended sequence of tests for patients with elevated cholesterol and triglycerides:

1. **Lipoprotein analysis** (lipid panel) after a 12 hour fast to determine levels of total serum cholesterol, TG, HDL cholesterol and estimated LDL cholesterol.
2. **VLDL cholesterol quantitation** to calculate the VLDL cholesterol /TG ratio. A ratio of >0.3 is highly suggestive of broad beta disease. This test also includes **lipoprotein electrophoresis** to identify  $\beta$ -VLDL (broad beta band).
3. **Apo E genotype test** to identify the apo E2/E2 genotype. This genotype is diagnostic for type III hyperlipoproteinemia when associated with elevation of serum cholesterol and TG.

### Apo E Genotype Testing Services:

- Diagnostic testing for type III hyperlipoproteinemia
- Predisposition testing in relatives
- Detailed reports with genetic interpretation, recommendations, and education
- Genetic consultation by board certified genetic counselors and geneticists
- Referral to a collaborating lipid laboratory for special lipid testing (VLDL quantitation and lipoprotein electrophoresis)
- Clinical consultations regarding type III hyperlipoproteinemia and other dyslipidemias may be arranged

**Method:** Apo E genotype testing involves DNA analysis by polymerase chain reaction (PCR) with restriction enzyme cleavage.

**Accuracy:** >99%

### Specimen Requirements:

- 5 ml whole blood in an EDTA (lavender top) tube, room temperature.

### Turnaround Time:

- 1 business day

**Please call Kimball Genetics for more information.**

