Dysbetalipoproteinemia

WHAT YOU NEED TO KNOW

What is dysbetalipoproteinemia?

Dysbetalipoproteinemia (DBL) is a genetic condition that affects how the body clears fats from the blood, leading to high triglycerides and a buildup of remnant cholesterol, which increases the risk of heart disease. It occurs in people who inherit two copies of the E2 version of the APOE gene, which reduces the liver's ability to remove fats from the blood—though not everyone with this gene develops DBL, it is required for the condition to occur.

What is the Criteria for Dysbetalipoproteinemia?

- Dysbetalipoproteinemia (DBL) is identified by high cholesterol and high triglycerides.
- It often includes abnormal cholesterol-to–apoB ratios.
- One diagnostic approach uses:
  - Total cholesterol / apoB > 6.2
  - Triglycerides / apoB < 10
- Different diagnostic methods may give different results.

How is Dysbetalipoproteinemia Diagnosed?

- Suspect dysbetalipoproteinemia (DBL) when both cholesterol and triglycerides are high, especially if apoB is low.
- Physical signs may include:
  - Yellow-orange discoloration on palm creases (palmar xanthomas)
  - Small yellow bumps on elbows or knees (eruptive xanthomas)
- Diagnosis can be confirmed by:
  - Genetic testing showing APOE E2/E2 genotype
- Specialized blood tests can detect abnormal lipoproteins, but these tests are rarely used.

What Lifestyle Modifications are Recommended for Patients?

- Restricting dietary cholesterol to <300 mg/day
- Limiting alcohol use
- Limiting carbohydrates to <40% of calories
- Avoiding simple carbohydrates
- Consume marine-derived omega-3 fatty acids
- Calorie restriction to promote weight loss

What Treatment Options are Available?

- If lifestyle changes are not enough, cholesterol-lowering medications are recommended for DBL.
- The most commonly used medications are:
  - Statins
  - Fibrates
- These medicines help lower high cholesterol and triglycerides.
- Other cholesterol-lowering treatments may also be options, though they are less studied in DBL, including:
  - Ezetimibe
  - Bempedoic acid
  - Icosapent ethyl
  - PCSK9 inhibitors
  - ApoC-III or ANGPTL3 inhibitors

To find a lipid specialist in your area, use the "**find a clinician**" tool on *learnyourlipids.com*.

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