

Casual Friday Series

# **Familial Hypercholesterolemia**

A Biogenetix Clinical Presentation

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# Disclaimer

- *Information in this presentation is not intended, in itself, to diagnose, treat, reverse, cure, or prevent any disease. While this presentation is based on medical literature, findings, and text, The following statements have not been evaluated by the FDA.*
- *The information provided in this presentation is for your consideration only as a practicing health care provider. Ultimately you are responsible for exercising professional judgment in the care of your own patients.*

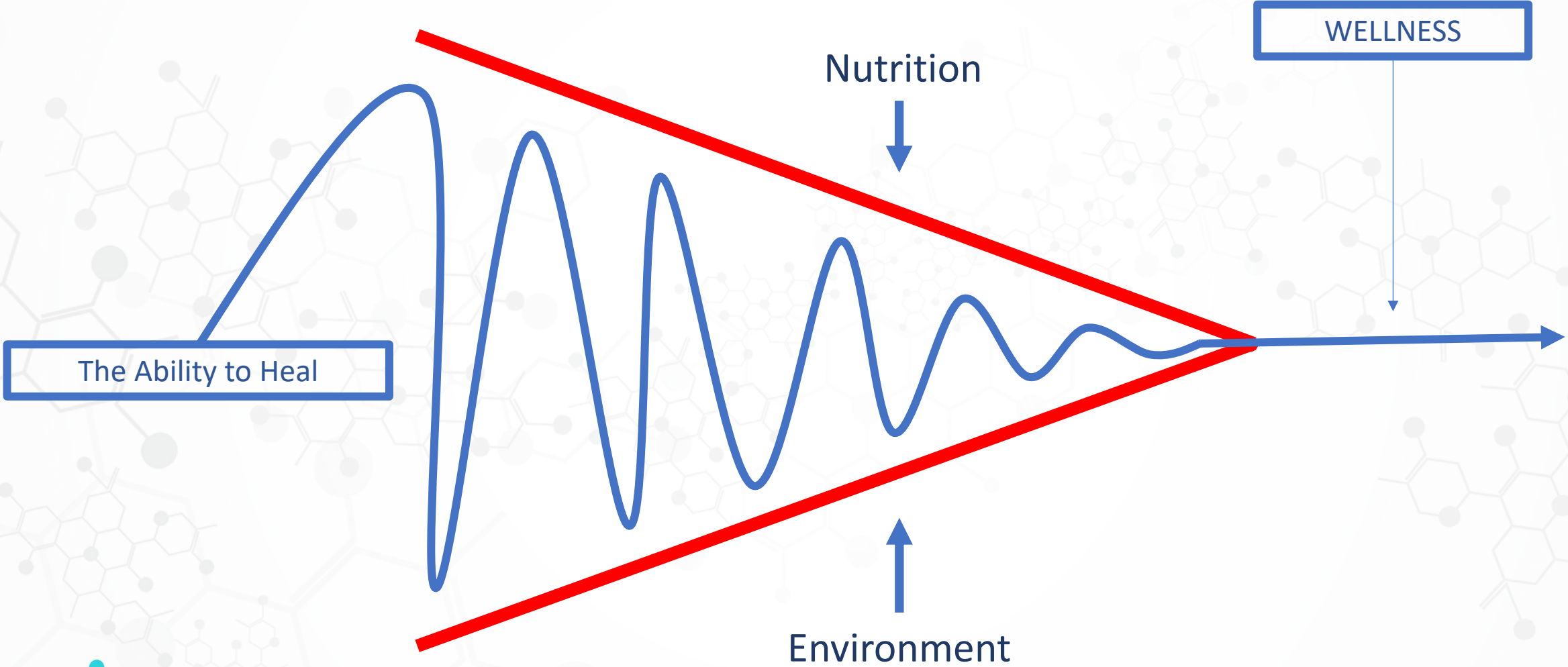




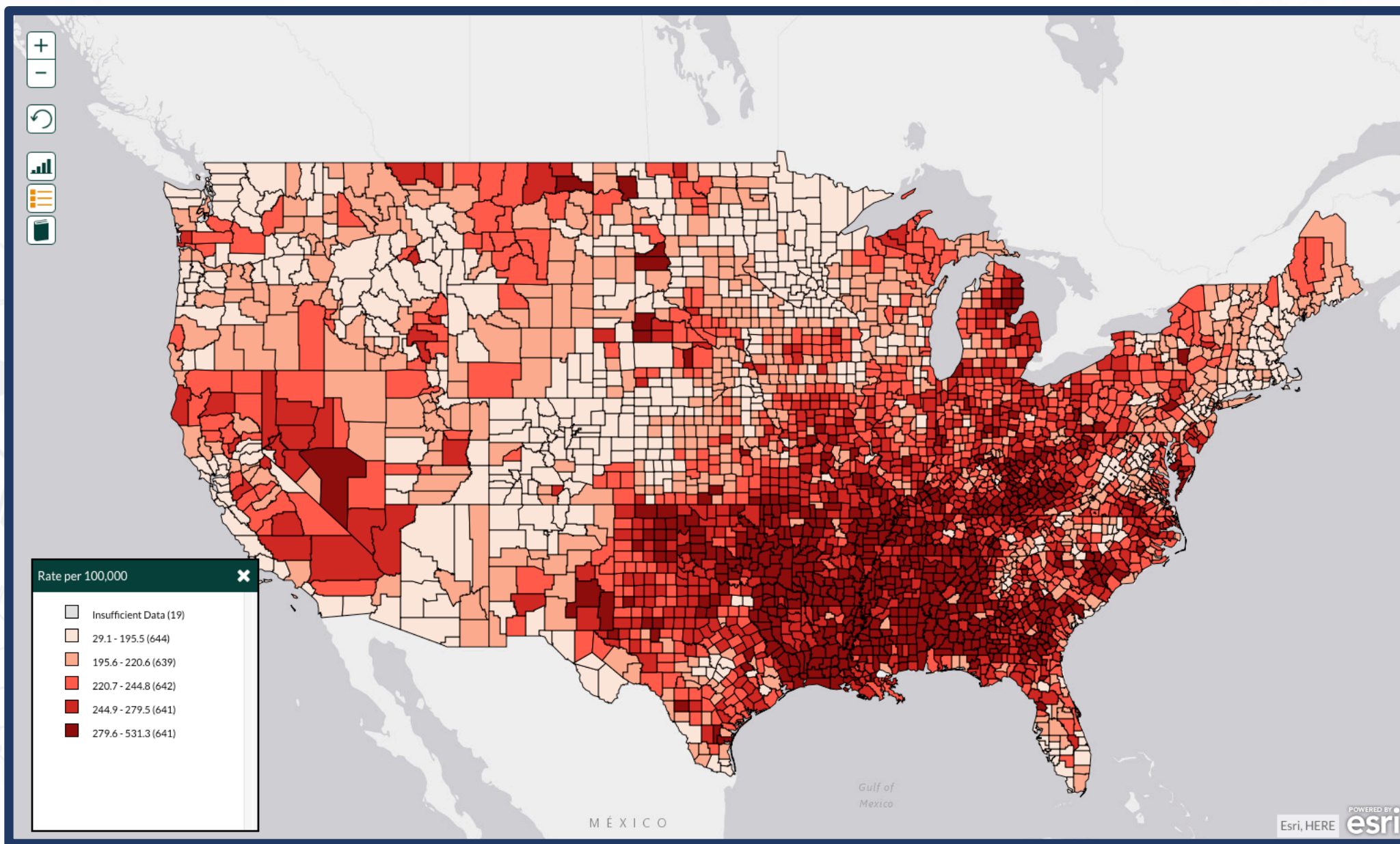
Lifestyle + Genetics = Chronic Health IMPROVEMENT



# Protocols







# NMR Lipoprofile

**Clinical Significance:** The Lipoprotein Fractionation NMR test is used to help assess the risk for cardiovascular disease (CVD) in patients with intermediate or high risk based on traditional or emerging risk factors, and to assess therapeutic response in patients undergoing lipid-lowering therapy, by quantification of the number and size of lipoprotein particles. The lipid panel is used, along with other test, during routine assessment to determine an individual's risk of cardiovascular disease. A lipid panel can also be used to monitor the efficacy of lifestyle interventions or medications.



## Beneficial Effects of Omega-3 Fatty Acids on Low Density Lipoprotein Particle Size in Patients with Type 2 Diabetes Already under Statin Therapy

Myung  
Youn

Beyond statin therapy for reducing low density lipoprotein cholesterol (LDL-C), additional therapeutic strategies are required to achieve more optimal reduction in cardiovascular risk among diabetic patients with dyslipidemia. To evaluate the effects and the safety of combined treatment with omega-3 fatty acids

There were no significant difference in the initial (week 0) lipid profiles among the three groups. After 8 weeks of treatment, as shown in [Table 1](#) and [Fig. 1](#), mean LDL particle size increased in all groups, and the percentage change was significantly greater in patients taking 4 g of omega-3 fatty acid with statin than in patients receiving statin monotherapy ( $2.8\% \pm 3.1\%$  vs.  $2.3\% \pm 3.6\%$ ,  $P=0.024$ ). Significant reduction in TG level was shown after 8-week treatment in all groups. The percentage change from baseline TG level was significantly greater in O3FA4S group than in the control group ( $-41.0\% \pm 24.1\%$  vs.  $-24.2\% \pm 31.9\%$ ,  $P=0.049$ ). TC level was significantly reduced at 8 weeks from baseline only in O3FA4S group ( $-0.44\% \pm 0.66$  mg/dL,  $P=0.018$ ), but the percentage change was not significantly different compared to the control group. In all groups, neither HDL-C nor LDL-C level showed any significant change during the study period. There were no significant differences between O3FA2S group and the control group after 8 weeks of respective treatment.







## Omega-3

### Supplement Facts

Serving Size: 1 Softgel  
Servings Per Container: 60

| Amount Per Serving             |        |  | %Daily Value |
|--------------------------------|--------|--|--------------|
| Calories                       | 10     |  |              |
| Total Fat                      | 1 g    |  | 1%†          |
| MaxSimil® Fish Oil Concentrate | 1.3 g  |  | **           |
| Total Omega-3 Fatty Acids      | 860 mg |  | **           |
| EPA (eicosapentaenoic acid)    | 600 mg |  | **           |
| DHA (docosahexaenoic acid)     | 260 mg |  | **           |

† Percent Daily Values are based on a 2,000 calorie diet.

\*\* Daily Value not established.

**Other Ingredients:** Softgel (fish gelatin, vegetable glycerin, and purified water), GRAS enteric coating (ethylcellulose, sodium alginate, purified water, medium-chain triglycerides, oleic acid, vegetable stearic acid, and ammonium hydroxide), and mixed natural tocopherols.

**Contains:** Fish (anchovy and/or sardine [sources of fish oil], tilapia and/or pangasius [sources of fish gelatin]).

Manufactured using MaxSimil® fish oil. MaxSimil® is a registered trademark of Ingenutra Inc. Protected under US patents 8,119,690 and 8,198,324; Canadian patents 2672513 and 2677670.



1. [Promethease.com](https://www.promethease.com)
2. [Strategene.me](https://www.strategene.me)
3. [SNPedia.com](https://www.snppedia.com)



# Familial Hypercholesterolemia

The *APOB* gene provides instructions for making two versions of the apolipoprotein B protein, a short version called apolipoprotein B-48 and a longer version known as apolipoprotein B-100. Both of these proteins are components of lipoproteins, which are particles that carry fats and fat-like substances (such as cholesterol) in the blood.

B-48 is gut based and associated with chylomicrons and dietary fat absorption.

B-100 is liver based and a building block to LDL *particles*.



# Familial Hypercholesterolemia

Most people with familial hypercholesterolemia inherit one altered copy of the *APOB* gene from an affected parent and one normal copy of the gene from the other parent. These cases are associated with an increased risk of early heart disease, typically beginning in a person's forties or fifties. Rarely, a person with familial hypercholesterolemia is born with two mutated copies of the *APOB* gene. This situation occurs when the person has two affected parents, each of whom passes on one altered copy of the gene. The presence of two *APOB* gene mutations results in a more severe form of hypercholesterolemia that usually appears in childhood.





# Familial Hypercholesterolemia

The APOB gene provides instructions for making a protein called apolipoprotein B. This protein helps LDL cholesterol bind to LDL receptors on the surface of cells, particularly in the liver. Certain variants in this gene reduce the ability of LDL cholesterol to bind to its receptor, causing fewer LDL cholesterol particles to be removed from the blood.



# Familial Hypercholesterolemia

B-48



Chylomicrons,  
gut level.

B-100



Repackaged in the liver with  
B-100 to form trig rich VLDL,  
and chol rich LDL.



Membrane health, sex  
hormones, steroids, etc.



# Familial Hypercholesterolemia

B-48



Chylomicrons,  
gut level.

B-100



Repackaged in the liver with  
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Membrane health, sex  
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# Familial Hypercholesterolemia

1. APOB *gene* codes for part of the particle.
2. Apolipoprotein B is a reflection of particle number.

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# Familial Hypercholesterolemia

B-48



Chylomicrons,  
gut level.

B-100



Produced in the liver with  
B-100 and high VLDL  
and cholesterol



Membrane health, sex  
hormones, steroids, etc



# Report Time!



1. [Promethease.com](https://www.promethease.com)
2. [Strategene.me](https://www.strategene.me)
3. [SNPedia.com](https://www.snppedia.com)



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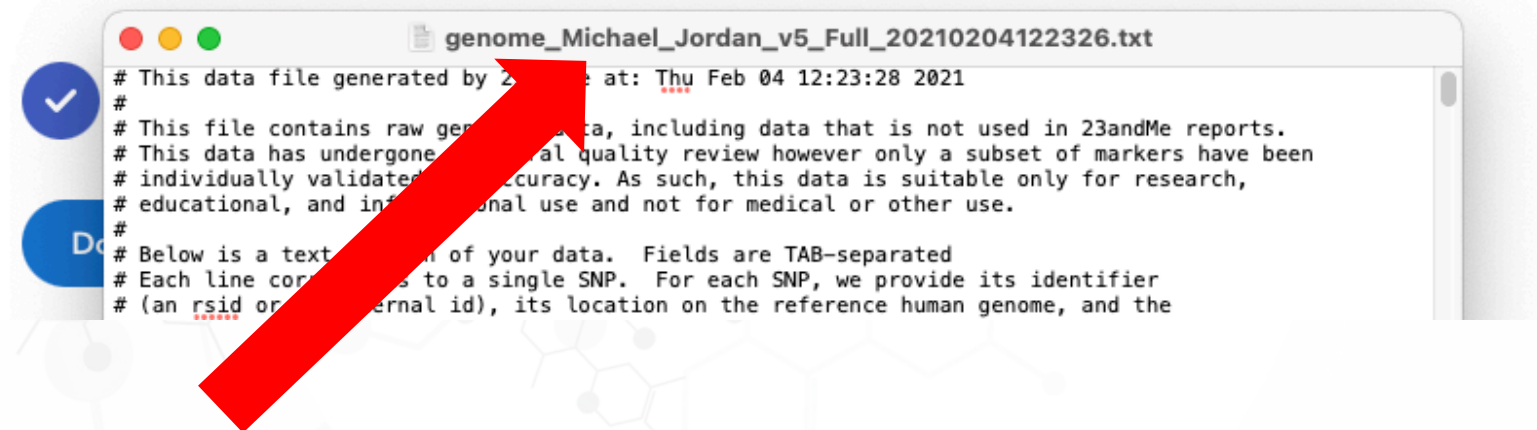
As part of your 23andMe service, we provide full access to your raw genetic data and do with as you please. Before downloading, however, you should read the terms and conditions that you should know about.



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## rs35705950(G;T)

**Possible miscall in Ancestry v2d data; otherwise, moderately (>5x) higher risk for lung issues (fibrosis or pneumonia)** See discussion via main rs-page.

rs35705950 is a SNP located 3 kb upstream of the start of transcription from the mucin 5B MUC5B gene. Two independent research groups both published articles in April 2011 associating the minor (T) allele of rs35705950 with highly increased risk for interstitial pneumonia and/or pulmonary fibrosis. For interstitial pneumonia, the odds ratio for heterozygotes was 6.8 (CI: 3.9 - 12.0) and for homozygotes, 20.8 (CI: 3.8 - 113.7). For pulmonary fibrosis, the reported odds ratios for heterozygotes and homozygotes in one study were 9.0 (CI: 6.2 to 13.1) and 21.8 (CI: 5.1 to 93.5), respectively, and were 5.9 (CI: 4.4 - 7.8) and 9.7 (CI: 4.7 - 19.9) in the other.

[more info](#)

|            |                      |
|------------|----------------------|
| Bad        | Repute               |
| 4.4        | Magnitude            |
| 2019-01-02 | Geno Modified        |
| 0.05234    | GMAF                 |
| Other      | ClinVar Significance |
| 16         | Publications         |
| 11         | Chromosome           |
| 1219991    | Position             |
| 5.8        | Max Magnitude        |
| 2019-12-08 | Rs Modified          |
| plus       | Stabilized           |
| plus       | Orientation          |

Medical Conditions Pulmonary fibrosis

ClinVar Other Idiopathic fibrosing alveolitis [[Pulmonary fibrosis]]

## gs144



Male Male.

|            |               |
|------------|---------------|
| 4          | Magnitude     |
| 2015-01-07 | Geno modified |

All ClinVar

Topics

Medical Conditions

ClinVar Diseases

Genes

Sort by Magnitude

CEU - Caucasians from Northern an...

Show: ☒ Genosets ☒ Homozyg  
☒ Heterozyg

Repute: ☒ Good ☒ Not Set ☒ Bad

Magnitude:  
- + 0 4.4 - +

Publications:  
- + 0 234 - +

Frequency:  
- + 0 100 - +

Require: ☐ Frequency

UI: ☒ Tooltips ☐ Editor Mode  
- + Font Size

Allow 10 2x more

Visible 10

Offscreen 23526

Good Not Set  
Bad



Vision: Normal



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Medical Conditions Pulmonary fibrosis

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Male Male.

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| 2015-01-07 | Geno modified |



Show: ☒ Genosets ☒ Homozyg  
☒ Heterozyg

Repute: ☐ Good ☐ Not Set ☒ Bad

Magnitude:

- + 0 4.4 - +

Publications:

- + 0 234 - +

Frequency:

- + 0 100 - +

Require: ☐ Frequency

UI: ☒ Tooltips ☐ Editor Mode

- + Font Size

Allow

227

2x more

Visible

10

Offscreen

227

☒ Good ☐ Not Set  
☒ Bad



Vision: Normal





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| 11         | Chromosome           |
| 1219991    | Position             |
| 5.8        | Max Magnitude        |
| 2019-12-08 | Rs Modified          |
| plus       | Stabilized           |
| plus       | Orientation          |

Medical Conditions Pulmonary fibrosis

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## gs144



Male Male.

|            |               |
|------------|---------------|
| 4          | Magnitude     |
| 2015-01-07 | Geno modified |



- Medical Conditions
- 3-Methylglutaconic aciduria type 2 (1)
- ABO blood group (11)
- AIDS (10)
- Abdominal aortic aneurysm (7)
- Achondroplasia (1)
- Acute lymphoblastic leukemia (4)
- Acute myeloid leukemia (1)
- Addiction (3)
- Age related macular degeneration

- + 0 234 - +

- + 0 100 - +

- + Font Size

227

2x more

227

10

Good Not Set  
Bad



rs1333049(C;C)

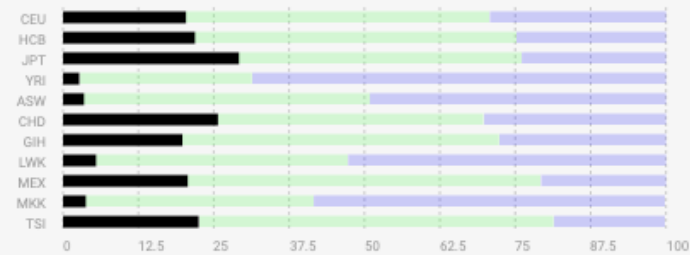
1.9x increased risk for coronary artery disease

rs1333049 has been reported in a large study to be associated with heart disease, in particular, coronary artery disease. The risk allele (oriented to the dbSNP entry) is most likely (C); the odds ratio associated with heterozygotes is 1.47 (CI 1.27-1.70), and for homozygotes, 1.9 (CI 1.61-2.24). This SNP has also been reported to have the highest association of any SNP studied in a subsequent experiment conducted with the resources of the German MI [Myocardial Infarction] Family Study. [ ] The initial studies were conducted on Caucasian populations. A subsequent study of Japanese and Korean patients has found rs1333049 to be associated with increased coronary artery disease risk, with roughly similar odds ratios. **Further reading (with comments)** - A long-term study of a cohort of 769 individuals finds ...

- Shared genetic susceptibility to ischemic stroke and coronary artery disease: a genome-wide analysis of common variants.
- Genome-wide association study for coronary artery calcification with follow-up in myocardial infarction.
- Two-marker association tests yield new disease associations for coronary artery disease and hypertension.
- A Genome-wide Association Study Identifies LIPA as a Susceptibility Gene for Coronary Artery Disease.
- Genomewide association analysis of coronary artery disease.
- Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls....

[more info](#)

|       |           |
|-------|-----------|
| Bad   | Repute    |
| 4     | Magnitude |
| 20.4% | Frequency |



|            |               |
|------------|---------------|
| 2016-12-20 | Geno Modified |
| 0.4334     | GMAF          |
| 101        | Publications  |
| 9          | Chromosome    |
| 22125504   | Position      |
| 4          | Max Magnitude |
| 2019-12-15 | Rs Modified   |
| plus       | Stabilized    |
| plus       | Orientation   |





# Familial Hypercholesterolemia

B-48



Chylomicrons,  
gut level.

B-100



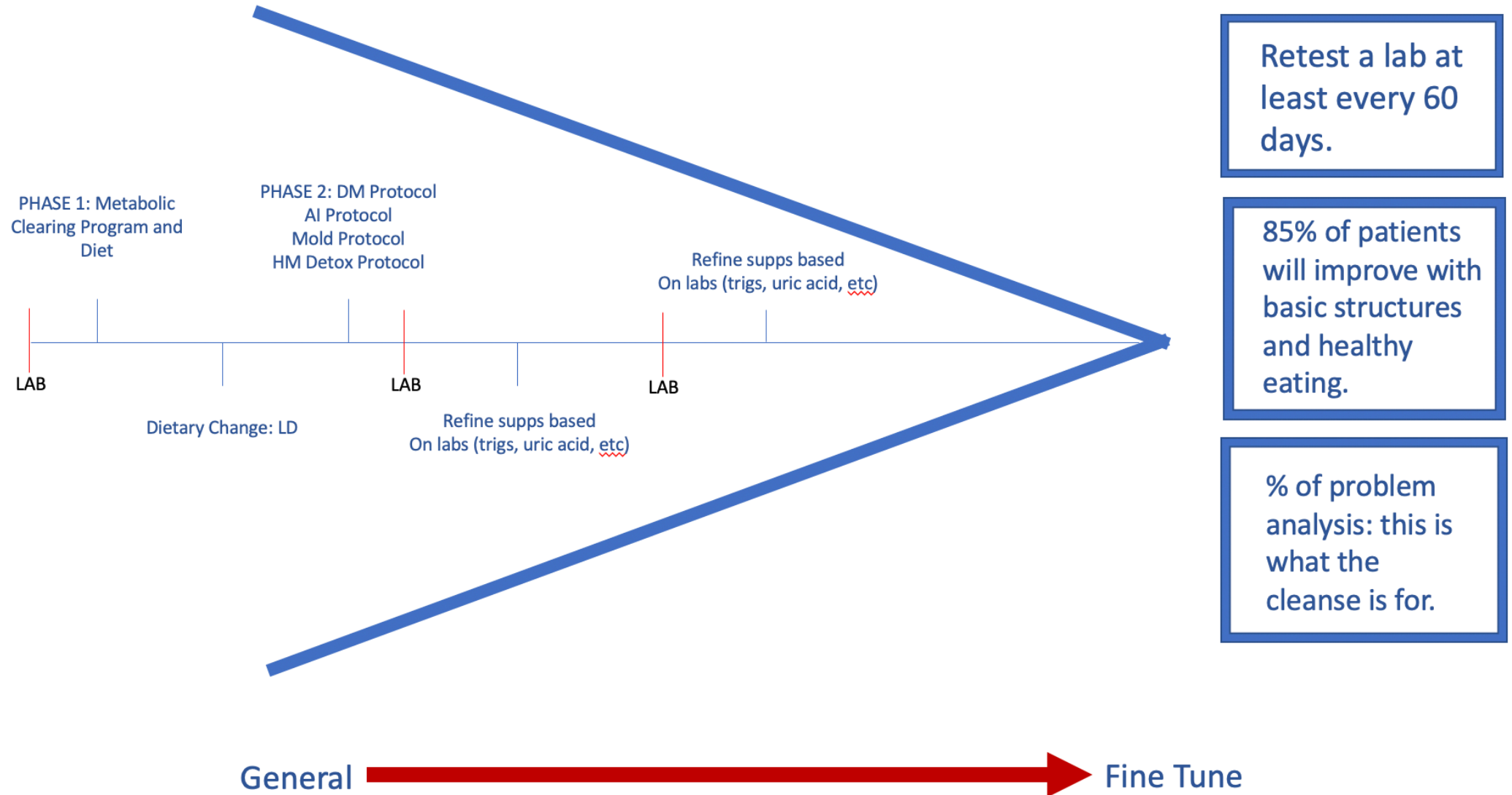
Produced in the liver with  
B-100 and high VLDL  
and cholesterol



Membrane health, sex  
hormones, steroids, etc



## Supplement and Diet Protocols



# Biogenetix: 833-525-0001



[zeb@biogenetix.com](mailto:zeb@biogenetix.com)



[kim@biogenetix.com](mailto:kim@biogenetix.com)

