

## **SUPPLEMENTAL MATERIAL**

### **Low prevalence of mutations in known loci for autosomal dominant hypercholesterolemia in a multi-ethnic patient cohort**

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**Table S1.** Lipid and lipoprotein profile on lipid lowering therapy <sup>g</sup>

|  | FH ( <i>LDLR</i> ) Patients |              | Unexplained ADH Patients |              | Group x Gender Interaction p-value |
|--|-----------------------------|--------------|--------------------------|--------------|------------------------------------|
|  | Male                        | Female       | Male                     | Female       |                                    |
| n  | 11                          | 19           | 25                       | 28           |                                    |
| Total Cholesterol (mg/dl)                  | 204 (26)                    | 215 (36)     | 182 (45) <sup>d</sup>    | 207 (40)     | 0.44                               |
| LDL-C (mg/dl)                              | 145 (26) <sup>a</sup>       | 135 (34)     | 116 (36)                 | 123 (31)     | 0.28                               |
| TG (mg/dl)                                 | 137 (57)                    | 140 (68)     | 127 (55) <sup>d</sup>    | 162 (51)     | 0.17                               |
| HDL-C (mg/dl)                              | 39.4 (6.9) <sup>d</sup>     | 51.8 (10.8)  | 43.7 (8.6) <sup>e</sup>  | 55.4 (18.0)  | 0.90                               |
| % LDL-C change from untreated value        | -48.5 (13.3)                | -53.1 (13.0) | -52.2 (14.9)             | -48.3 (10.8) |                                    |
| Absolute LDL-C change from untreated value | -143 (65)                   | -159 (55)    | -130 (51)                | -116 (36)    |                                    |

Continuous variables are summarized as mean (SD).

P-values are from two-way ANOVA for continuous variables and the Breslow-Day test for categorical variables.

<sup>a</sup> p<0.05, FH (*LDLR*) versus unexplained ADH for the same gender.

<sup>d</sup> p<0.05, <sup>e</sup> p<0.01, Male versus Female for the same group

<sup>g</sup> Includes statins, ezetemibe, bile acid sequestrants, niacin, fish oil, and/or sitostanol.

**Table S2.** Percentage of patients on lipid lowering medication

|   | FH ( <i>LDLR</i> ) Patients |        | Unexplained ADH Patients |        | Median daily dose (range) |
|---|-----------------------------|--------|--------------------------|--------|---------------------------|
|   | Male                        | Female | Male                     | Female |                           |
| N   | 11                          | 19     | 25                       | 28     |                           |
| Lipid lowering medication                       |                             |        |                          |        |                           |
| Statin (%)                                      | 90.9                        | 94.7   | 100.0                    | 82.1   |                           |
| Rosuvastatin (%)                                | 54.6                        | 68.4   | 76.0                     | 35.7   | 40 mg<br>(5 – 40)         |
| Simvastatin (%)                                 | 18.2                        | 21.1   | 16.0                     | 21.4   | 40 mg<br>(10 – 80)        |
| Atorvastatin (%)                                | 9.1                         | 5.3    | 4.0                      | 10.7   | 60 mg<br>(10 – 80)        |
| Pravastatin (%)                                 | 9.1                         | 0      | 8.0                      | 14.3   | 20 mg<br>(10 – 40)        |
| Ezetimibe (%)                                   | 63.6                        | 52.6   | 44.0                     | 35.7   | 10 mg<br>(5 – 10)         |
| Niacin (%)                                      | 0.0                         | 0.0    | 4.0                      | 3.6    | 1000 mg<br>(500 – 1500)   |
| Bile acid sequestrants* (%)                     | 27.3                        | 10.5   | 8.0                      | 21.4   |                           |
| Fish oil (%)                                    | 9.1                         | 0.0    | 12.0                     | 10.7   | 2 g<br>(2 – 3)            |
| Lipid lowering medication class                 |                             |        |                          |        |                           |
| Statin alone (%)                                | 36.4                        | 42.1   | 52.0                     | 50.0   |                           |
| Ezetimibe alone (%)                             | 0                           | 5.3    | 0                        | 10.7   |                           |
| Bile acid sequestrants alone (%)                | 0                           | 0      | 0                        | 3.6    |                           |
| Statin + Ezetimibe (%)                          | 36.4                        | 42.1   | 40.0                     | 17.9   |                           |
| Statin + bile acid sequestrants (%)             | 0                           | 5.3    | 4.0                      | 10.7   |                           |
| Ezetimibe + bile acid sequestrants (%)          | 9.1                         | 0      | 0                        | 3.6    |                           |
| Statin + Ezetimibe + bile acid sequestrants (%) | 18.2                        | 5.3    | 4.0                      | 3.6    |                           |

\* Colestipol, cholestyramine, and colesevelam were used