**Tablas y anexos**

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| --- | --- | --- |
| Tabla 1 | | |
| Características de los participantes | | **n=36** |
| Edad-años | |  |
|  | Media | 27 |
|  | DE | 17 |
| Sexo |  |  |
|  | Mujer | 66 % (24) |
|  | Hombre | 34 % (12) |
| Región | |  |
|  | Costa Norte | 10 |
|  | Antioquia y Eje Cafetero | 8 |
|  | Valle del Cauca | 8 |
|  | Santander | 6 |
|  | Bogotá | 4 |

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| **Tabla 2**  **Características clínicas** | |  |
|  |  | n=36 |
| **Puntaje RCLH** | |  |
|  | Definitivo HF (> 8 puntos) | 94,4 % (34) |
|  | Probable HF (6-8 puntos) | 5,6 % (2) |

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| **Tabla 3**  **Tratamiento hipolipemiante** | **n** | **%** | **Dosis media** |
| Rosuvastatina | 24 | 67 % |  |
| Menor de 18 años | 9 |  | 22,2 mg |
| Mayor o igual a 18 años | 15 |  | 33,3 mg |
| Atorvastatina | 8 | 22 % |  |
| Menor de 18 años | 5 |  | 20 mg |
| Mayor o igual a 18 años | 5 |  | 80 mg |
| Lovastatina | 1 | 3 % | 20 mg |
| Simvastatina | 1 | 3 % | 40 mg |
| Ezetimibe | 30 | 83 % | 10 mg |
| Lomitapide | 20 | 56 % | 12,7 mg |
| Evolocumab | 2 | 6 % | 420 mg |

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| --- | --- | --- | --- |
| Tabla 4  Valores basales y finales de lípidos, y cambio en porcentaje | | | |
|  | Niveles basales | Niveles finales | Cambio |
| TC (mg/dl) | 607,3 | 305,9 | 50% |
| LDL-C (mg/dl) | 533,7 | 245,1 | 54% |
| Estatina más ezetimibe sin lomitapide (n=11) | 487,4 | 319,1 | 35% |
| Estatina más ezetimibe más lomitapide (n=18) | 602,0 | 190,5 | 68 % |
| Estatina y ezetimibe, o ambas, más evolucumab (n=2) | 439,6 | 413,5 | 5 % |
| Estatina sola (n=1) | 299 | 183 | 38 % |
| HDL (mg/dl) | 40,7 | 47,7 | 17 % |
| TG (mg/dl) | 142,4 | 115,4 | 19 % |

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| **Tabla 5**  **Hallazgos genéticos** | | **n=34** |
|  |  | % (n) |
| **Homocigótico simple** | | 38,9 % (14) |
|  | Edad (media) | 22,5 |
|  | Mujeres | 42 %(6) |
|  | Región Costa Norte | 50 %(7) |
| **Heterocigótico compuesto** | | 33,3 % (12) |
|  | Edad (media) | 24,2 |
|  | Mujeres | 66,6 (8) |
|  | Región Santander | 25 % (3) |
|  | Región Eje Cafetero | 25 % (3) |
| **Heterocigótico doble** | | 5,6 % (2) |
|  | Edad (media) | 44,5 |
|  | Hombres | 100 % (2) |
|  | Región Antioquia | 50 % (1) |
| **Autosómico recesivo** | | 2,8 % (1) |
|  | Edad (media) | 8 |
|  | Hombres | 100 % (1) |
|  | Región Antioquia | 100 % (1) |
| **Heterocigótico simple** | | 13,9 % (5) |
|  | Edad (media) | 29,4 |
|  | Hombres | 100 % (1) |
|  | Región Valle | 40 % (2) |

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| **Tabla 6**  **Característica** | **Proteína** | **Mutación** | **N** |
| **Homocigótico simple** | LDLR | c.11G>A | 8 |
|  |  | c.314-?\_940+?del | 1 |
|  |  | c.314-1121\_941-1446del | 2 |
|  |  | c.1587-?\_1845+?del | 1 |
|  |  | c.682G>A | 1 |
|  |  | c.1878delA | 1 |
| **Homocigótico compuesto** | LDLR | c.502G>A/c.1358+2T>G | 1 |
|  |  | c.502G>A/c.682G>A | 1 |
|  |  | c.1878.del/c.190+4A>T | 1 |
|  |  | c.11G>A/c.1816G>A | 1 |
|  |  | c.81C>G/c.809G>A | 1 |
|  |  | c.11G>A/c.622>A | 1 |
|  |  | c.1187-1G>T/c.-135C>G | 1 |
|  |  | c.68-?\_940+?del | 1 |
|  |  | c.1187-1G>T- c.1587-?\_1845+?de | 2 |
|  |  | c.1187-1G>T/c.2479G>A | 1 |
|  |  | c.314-?\_940+?del/c.1246C>T | 1 |
| **Heterocigótico doble** | LDLR y APOB | c.11G>A/c.13369.G>A | 1 |
|  | LDLR y APOE | c.682G>A/c.526C>T. | 1 |
| **Autosómico recesivo** | LDLRAP1 | c.88+5G>A | 1 |
| **Heterocigótico simple** | LDLR | c.11G>A | 1 |
|  |  | c.1216C>A | 1 |
|  |  | c.11G>A | 1 |
|  |  | c.314-?\_940+?del | 2 |
|  |  | **Total** | 34 |

**Tabla 7** Características demográficas y clínicas de los pacientes con HF-Ho

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **ID** | **Región** | **Edad (años)** | **Género** | **Puntaje RCLH** | **Genotipo** | **Proteína** | **Xantoma** | **Arco corneal** | **Xantelasma** | **ACV** | **Estenosis carotídea** | **Estenosis aórtica** | **Enfermedad coronaria** |
| 1 | Antioquia | 11 | M | >8 | HF-Ho S | LDLR | + | + | + | - | - | + | - |
| 2 | Antioquia | 11 | H | >8 | HF-Ho S | LDLR | + | + | - | - | - | + | - |
| 3 | Sucre | 33 | M | >8 | HF-Ho S | LDLR | + | + | + | - | + | + | + |
| 4 | Sucre | 18 | M | >8 | HF-Ho S | LDLR | + | - | - | - | + | + | + |
| 5 | Sucre | 22 | H | >8 | HF-Ho S | LDLR | + | - | - | - | - | + | + |
| 6 | Sucre | 5 | H | >8 | HF-Ho S | LDLR | + | + | - | - | + | - | + |
| 7 | Sucre | 13 | H | >8 | HF-Ho S | LDLR | + | - | - | - | - | - | - |
| 8 | Cesar | 13 | H | >8 | HF-Ho S | LDLR | + | + | - | - | - | - | - |
| 9 | Valle | 23 | H | >8 | HF-Ho S | LDLR | + | + | - | - | - | + | - |
| 10 | Valle | 43 | M | >8 | HF-Ho S | LDLR | + | + | - | - | - | - | + |
| 11 | Valle | 56 | M | >8 | HF-Ho S | LDLR | + | + | + | - | + | - | + |
| 12 | Sucre | 27 | H | >8 | HF-Ho S | LDLR | + | + | + | - | + | - | - |
| 13 | Santander | 23 | M | >8 | HF-Ho S | LDLR | + | - | - | - | - | - | - |
| 14 | Valle | 18 | H | >8 | HF-Ho S | LDLR | - | - | - | - | - | - | - |
| 15 | Santander | 14 | H | >8 | HF-Ho C | LDLR | + | + | - | - | - | - | - |
| 16 | Santander | 44 | H | >8 | HF-Ho C | LDLR | + | + | - | - | + | - | + |
| 17 | Valle | 14 | M | >8 | HF-Ho C | LDLR | + | + | - | - | - | + | - |
| 18 | Caldas | 37 | M | >8 | HF-Ho C | LDLR | + | + | - | - | - | - | - |
| 19 | Santander | 12 | M | >8 | HF-Ho C | LDLR | + | - | - | - | - | - | - |
| 20 | Bogotá | 14 | H | >8 | HF-Ho C | LDLR | + | - | - | - | - | - | - |
| 21 | Bogotá | 11 | M | >8 | HF-Ho C | LDLR | + | + | - | - | + | + | - |
| 22 | Sucre | 4 | M | >8 | HF-Ho C | LDLR | + | - | - | - | - | - | - |
| 23 | Quindío | 53 | M | >8 | HF-Ho C | LDLR | - | + | + | - | - | + | + |
| 24 | Bogotá | 54 | M | >8 | HF-Ho C | LDLR | + | + | - | - | - | - | - |
| 25 | Quindío | 7 | H | >8 | HF-Ho C | LDLR | + | - | - | - | - | - | - |
| 26 | Valle | 27 | M | >8 | HF-Ho C | LDLR | + | - | - | - | - | - | - |
| 27 | Antioquia | 35 | M | >8 | HF- He D | LDLR y APOB | - | + | - | - | - | - | - |
| 28 | Santander | 54 | M | >8 | HF- He D | LDLR y APO E | - | + | + | - | - | - | + |
| 29 | Antioquia | 8 | M | >8 | AR | LDLRAP1 | + | - | + | - | - | - | - |
| 30 | Córdoba | 27 | M | >8 | HF-He S | LDLR | + | - | - | - | - | - | + |
| 31 | Bogotá | 56 | M | >8 | HF-He S | LDLR | - | - | - | - | + | - | + |
| 32 | Antioquia | 46 | M | >8 | HF-He S | LDLR | - | + | - | - | - | - | + |
| 33 | Valle | 4 | M | 6-7 | HF-He S | LDLR | - | - | - | - | - | - | - |
| 34 | Valle | 14 | M | 6-7 | HF-He S | LDLR | - | - | - | - | - | - | - |
| 35 | Santander | 53 | M | >8 | No autorizó | | - | - | - | + | - | - | + |
| 36 | Sucre | 57 | M | >8 | No autorizó | | - | - | - | + | - | - | + |

HF-Ho S: homocigótico simple, HF-Ho C: heterocigoto compuesto, HF-Ho D: heterocigótico doble. HF-He S: heterocigótico simple.

**Tabla 8** Genotipificación de los alelos de los pacientes con HF-Ho

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
|  | **Alelo 1** |  |  | **Alelo 2** | | |
| **ID** | **Gen** | **Mutación** | **Efecto** | **Gen** | **Mutación** | **Efecto** |
| 1 | LDLR | c.11G>A | Patogénica | LDLR | c.11G>A | Patogénica |
| 2 | LDLR | c.11G>A | Patogénica | LDLR | c.11G>A | Patogénica |
| 3 | LDLR | c.11G>A | Patogénica | LDLR | c.11G>A | Patogénica |
| 4 | LDLR | c.11G>A | Patogénica | LDLR | c.11G>A | Patogénica |
| 5 | LDLR | c.11G>A | Patogénica | LDLR | c.11G>A | Patogénica |
| 6 | LDLR | c.11G>A | Patogénica | LDLR | c.11G>A | Patogénica |
| 7 | LDLR | c.11G>A | Patogénica | LDLR | c.11G>A | Patogénica |
| 8 | LDLR | c.11G>A | Patogénica | LDLR | c.11G>A | Patogénica |
| 9 | LDLR | c.314-?\_940+?del | Patogénica | LDLR | c.314-?\_940+?del | Patogénica |
| 10 | LDLR | c.314-1121\_941-1446del | Patogénica | LDLR | c.314-1121\_941-1446del | Patogénica |
| 11 | LDLR | c.314-1121\_941-1446del | Patogénica | LDLR | c.314-1121\_941-1446del | Patogénica |
| 12 | LDLR | c.1587-?\_1845+?del | Patogénica | LDLR | c.1587-?\_1845+?del | Patogénica |
| 13 | LDLR | c.682G>A | Patogénica | LDLR | c.682G>A | Patogénica |
| 14 | LDLR | c.1878delA | Patogénica | LDLR | c.1878delA | Patogénica |
| 15 | LDLR | c.502G>A | Patogénica | LDLR | c.1358+2T>G | Probablemente patogénica |
| 16 | LDLR | c.502G>A | Patogénica | LDLR | c.682G>A | Patogénica |
| 17 | LDLR | c.1878.del | Patogénica | LDLR | c.190+4A>T | Patogénica |
| 18 | LDLR | c.11G>A | Patogénica | LDLR | c.1816G>A | Variante de significado clínico incierto |
| 19 | LDLR | c.81C>G | Patogénica | LDLR | c.809G>A | Probablemente patogénica |
| 20 | LDLR | c.11G>A | Patogénica | LDLR | c.622>A | Patogénica |
| 21 | LDLR | c.1187-1G>T | Patogénica | LDLR | c.-135C>G | Probablemente patogénica |
| 22 | LDLR | c.68-?\_940+?del | Patogénica | LDLR | c.1587-?\_1845+?de | Patogénica |
| 23 | LDLR | c.1187-1G>T | Patogénica | LDLR | c.1187G>A | Variante de significado clínico incierto |
| 24 | LDLR | c.1187-1G>T | Patogénica | LDLR | c.1187G>A | Variante de significado clínico incierto |
| 25 | LDLR | c.1187-1G>T | Patogénica | LDLR | c.2479G>A | Probablemente patogénica |
| 26 | LDLR | c.314-?\_940+?del | Patogénica | LDLR | c.1246C>T | Patogénica |
| 27 | LDLR | c.11G>A | Patogénica | APOB | c.13369.G>A | Patogénica |
| 28 | LDLR | c.682G>A | Patogénica | APOE | c.526C>T. | Probablemente patogénica |
| 29 | LDLRAP1 | c.88+5G>A | Probablemente patogénica | LDLRAP1 | c.88+5G>A | Probablemente patogénica |
| 30 | LDLR | c.11G>A | Patogénica | LDLR | c.345C>G | ND |
| 31 | LDLR | c.1216C>A | Patogénica | ND | ND | ND |
| 32 | LDLR | c.11G>A | Patogénica | ND | ND | ND |
| 33 | LDLR | c.314-?\_940+?del | Patogénica | ND | ND | ND |
| 34 | LDLR | c.314-?\_940+?del | Patogénica | ND | ND | ND |

ND: no definida.