|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| PatientHOXA1 genotype | Age at diagnosis, yr | Sex | BAV Subtype | Predominant AV function | Family history | Leaflet calcification |
| **BAV30**-1His, WTA | 20.3 | M | Type 1, RC/LC | Moderate AR | no | - |
| **BAV32**+1**HisArg**, WTA | 24 | M | Type 0, ap | severe AR | no | - |
| **BAV51**-1His, WTA | 61 | M | Type 1, RC/LC | Severe AS | yes | + |
| **BAV58**+1His, WTA | 41 | F | Type 1, RC/LC | moderate AR | no | - |
| **BAV89**-1His, WTA | 68 | F | Type 1, RC/LC | Severe AS | no | +++ |
| **BAV103**-1His, WTA | 44 | F | Type 1; RC/LC | Small AR | no | - |
| **BAV116**+1**His**, WTA | 65 | F | Type 1, RC/LC | moderate AR | no | - |
| **BAV117**-1His, WTA | 68 | F | Type 1; RC/LC | Severe AS | no | ++ |
| **BAV121**-1His, WTA | 32 | F | Type 0, RC/LC | Normal | no | - |
| **BAV141****-3HisArg**, WTA | 30 | M | Type 0, RC/LC | Severe AR | no | - |
| **BAV148**-1His, **WTG** | 45 | M | Type 0, RC/LC | Severe AR | no | - |
| **BAV162**-1His, WTA | 80 | M | Type 1, RC/LC | Severe AS | no | ++ |
| **BAV164**-1His, WTA | 51 | M | Type 0, CD/CG | Normal | no | - |
| **BAV166****-3HisArg**, **-3HisArg** | 45 | M | Type 0,  | Severe AS and Severe AR | no | ++ |
| **BAV186**-1His, **WTG** | 52 | M | Type 2 | Moderate AS | no | ++ |
| **BAV204**-1His, WTA | 73 | M | Type 1, RC/LC | severe AS | no | ++ |
| **BAV220**-1His, WTA | 55 | M | Type 1, RC/LC | Normal | no | - |
| **BAV240**-1His, WTA | 65 | F | Type 1, RC/LC | Moderate AR | no | - |
| **BAV277**-1His, WTA | 75 | M | Type 1, RC/LC | Severe AS | no | ++ |
| **BAV291**-1His, WTA | 65 | M | Type 1, RC/LC | Severe AS | no | - |
| **BAV292**-1His, WTA | 72 | F | Type 1, RC/LC | Small AR | no | - |
| **BAV302**-1His, WTA | 80 | M | Type 1, RC/LC | Severe AS | no | ++ |
| **BAV313**+1**His**, WTA | 67 | F | Type 1, RC/LC | Severe AS and Moderate AR | no | ++ |
| **BAV326**-1His, WTA | 59 | F | Type 1, RC/LC | Severe AS | no | ++ |
| **BAV330**-1His, WTA | 39 | F | Type 1, RC/LC | Small AR | no | - |
| **BAV347**+1**His**, WTA | 45 | M | Type 1, RC/NC | Normal | no | + |
| **BAV378**-1His, WTA | 47 | M | Typepe 1, RC/LC | Moderate AR | no | - |
| **BAV386**-1His, WTA | 31 | M | Type 1, RC/LC | Severe AR | no | - |
| **BAV390**-1His, WTA | 41 | M | Type 1, RC/NC | Severe AR | yes | - |

**Supplementary Table 1: Clinical description of BAV patients with *HOXA1* variations**

Number of histidines in stretch are indicated according to genotypes described in table 2. For example, WTA= c.218G>A; WTG= c.218G; -1His = c.[213\_215delCCA;218G>A].