**Supplementary Table 1: Numerical data on height, arm span, AS-H and AS/H and their SDS for the 33 individuals with heterozygous *ACAN* haploinsufficiency**

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| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **MALES** |  |  |  |  |  |  |  |  |  |
|  |  |  |  |  |  |  |  |  |  |
| Patient | Age | Height | SDS | Arm  | SDS | Arm span | SDS | Arm span  | SDS |
| number | (years) | (cm) | span (cm)  | divided by | minus |
|  |  |  |  | height | height (cm) |
|  |  |  |  |  |  |
| CC-5 | 5.50 | 99.2 | -2.57 | 105.0 | -2.90 | 1.06 | 2.99 | 5.8 | 2.53 |
| CC-9a | 5.87 | 105.5 | -3.24 | 104.4 | -2.22 | 0.99 | -0.45 | -1.1 | 0.09 |
| CC-14  | 6.42 | 103.7 | -2.34 | 105.0 | -2.22 | 1.01 | 0.69 | 1.3 | 0.94 |
| CC-3a | 7.16 | 102.2 | -2.68 | 108.0 | -1.88 | 1.06 | 2.63 | 5.8 | 2.29 |
| CC-9 | 8.42 | 111.3 | -2.57 | 110.7 | -2.45 | 0.99 | -0.40 | -0.6 | -0.01 |
| KI-A17 | 13.00 | 152.0 | -4.06 | 159.0 | -4.54 | 1.05 | 1.28 | 7.0 | 1.51 |
| KI-A20 | 14.00 | 155.4 | -4.54 | 164.5 | -2.76 | 1.06 | 1.68 | 9.1 | 1.91 |
| KI-A16 | 15.00 | 152.0 | -4.06 | 163.0 | -3.26 | 1.07 | 2.35 | 11.0 | 2.25 |
| KI-B1 | 15.08 | 153.4 | -3.79 | 158.5 | -3.75 | 1.03 | 0.49 | 5.1 | 0.70 |
| KI-A2 | 27.00 | 157.0 | -2.68 | 168.0 | -1.88 | 1.07 | 1.60 | 11.0 | 1.92 |
| KI-A21 | 36.00 | 167.0 | -1.55 | 169.0 | -1.77 | 1.01 | -0.63 | 2.0 | -0.31 |
| KI-A14 | 39.00 | 152.0 | -3.24 | 165.0 | -2.22 | 1.09 | 2.19 | 13.0 | 2.41 |
| KI-A15 | 39.00 | 160.0 | -2.34 | 165.0 | -2.22 | 1.03 | 0.11 | 5.0 | 0.43 |
| CC-14a | 48.00 | 155.4 | -2.86 | 160.0 | -2.79 | 1.03 | 0.05 | 4.6 | 0.33 |
| KI-A9 | 64.00 | 158.0 | -2.57 | 163.0 | -2.45 | 1.03 | 0.13 | 5.0 | 0.43 |
| KI-A8 | 68.00 | 158.0 | -2.57 | 159.0 | -2.90 | 1.01 | -0.84 | 1.0 | -0.56 |
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|  |  |  |  |  |  |  |  |  |  |
| **FEMALES** |  |  |  |  |  |  |  |  |  |
|  |  |  |  |  |  |  |  |  |  |
| Patient | Age | Height | SDS | Arm  | SDS | Arm span | SDS | Arm span  | SDS |
| number | (years) | (cm) | span (cm)  | divided by | minus |
|  |  |  |  | height | height (cm) |
|  |  |  |  |  |  |
| CC-6 |  4.58 | 95.6 | -4.27 | 90.5 | -7.35 | 0.95 | -1.67 | -5.10 | -0.86 |
| CC-16 | 5.66 | 100.0 | -3.74 | 98.5 | -3.12 | 0.99 | -0.15 | -1.50 | 0.39 |
| CC-15 | 6.00 | 95.7 | -4.22 | 92.5 | -3.97 | 0.97 | -0.97 | -3.20 | -0.26 |
| CC-11 | 7.83 | 110.1 | -4.05 | 114.0 | -2.62 | 1.04 | 1.67 | 3.90 | 1.97 |
| KI-A1 | 11.33 | 140.2 | -2.47 | 146.1 | -2.12 | 1.04 | 1.78 | 5.90 | 2.01 |
| CC-12 | 18.00 | 149.5 | -3.50 | 159.0 | -1.98 | 1.06 | 1.77 | 9.50 | 2.44 |
| CC-4 | 20.00 | 153.9 | -2.80 | 151.5 | -3.04 | 0.98 | -1.54 | -2.40 | -0.62 |
| CC-15a | 35.83 | 124.4 | -7.51 | 121.2 | -7.35 | 0.97 | -1.97 | -3.20 | -0.82 |
| CC-16a | 35.83 | 146.1 | -4.05 | 154.5 | -2.62 | 1.06 | 1.51 | 8.40 | 2.16 |
| KI-A19 | 37.00 | 168.0 | -0.55 | 173.0 | 0.01 | 1.03 | 0.35 | 5.00 | 1.29 |
| CC-8a | 38.00 | 138.2 | -2.86 | 146.5 | -3.75 | 1.06 | 1.62 | 8.30 | 2.13 |
| KI-A13 | 39.00 | 146.0 | -1.55 | 150.0 | -3.26 | 1.03 | 0.25 | 4.00 | 1.03 |
| KI-B2 | 46.83 | 143.0 | -4.30 | 153.5 | -2.76 | 1.07 | 2.18 | 10.50 | 2.70 |
| KI-A23 | 47.00 | 145.4 | -4.29 | 146.0 | -3.94 | 0.97 | 0.11 | 0.60 | -0.25 |
| KI-A11 | 52.00 | 156.0 | -0.55 | 158.0 | -2.12 | 1.01 | -0.35 | 2.00 | 0.51 |
| KI-A10 | 62.00 | 148.0 | -3.74 | 151.0 | -3.12 | 1.02 | -0.04 | 3.00 | 0.77 |
| KI-A4 | 81.00 | 145.0 | -4.22 | 145.0 | -3.97 | 1.00 | -0.89 | 0.00 | 0.00 |
|  |  |  |  |  |  |  |  |  |  |