

Supplement to Nair et al., Human Molecular Genetics, 1997

Selected characteristics of the study cohort

A. Pedigree data

| Family type | <u>No. of families</u> | | | <u>Members evaluated</u> | | | <u>Affected members</u> | | |
|--------------|------------------------|-----|-------|--------------------------|-----|-------|-------------------------|-----|-------|
| | U.S. | Ger | Total | U.S. | Ger | Total | U.S. | Ger | Total |
| Nuclear | 60 | 26 | 86 | 224 | 97 | 321 | 157 | 70 | 227 |
| Extended: | | | | | | | | | |
| 2-generation | 1 | 5 | 6 | 8 | 29 | 37 | 7 | 24 | 31 |
| 3-generation | 9 | 11 | 20 | 183 | 138 | 321 | 69 | 72 | 141 |
| 4-generation | 2 | 1 | 3 | 34 | 13 | 47 | 12 | 3 | 15 |
| Total | 72 | 43 | 115 | 449 | 277 | 726 | 245 | 169 | 414 |

Selected characteristics of the study cohort (continued)

B. Sib pair distribution in pedigrees

| Family Type | Sibships with 2 affected sibs | | | Sibships with 3 affected sibs | | | Sibships with 4 affected sibs | | | Sibships with 5 affected sibs | | |
|--|----------------------------------|-----|-------|----------------------------------|-----|-------|----------------------------------|-----|-------|----------------------------------|-----|-------|
| | U.S. | Ger | Total | U.S. | Ger | Total | U.S. | Ger | Total | U.S. | Ger | Total |
| Nuclear | 48 | 24 | 72 | 10 | 2 | 12 | 2 | 0 | 2 | 0 | 0 | 0 |
| Extended: | | | | | | | | | | | | |
| 2-generation | 3 | 8 | 11 | 0 | 2 | 2 | 0 | 0 | 0 | 0 | 0 | 0 |
| 3-generation | 17 | 13 | 30 | 5 | 8 | 13 | 1 | 0 | 1 | 0 | 0 | 0 |
| 4-generation | 1 | 1 | 2 | 0 | 0 | 0 | 0 | 0 | 0 | 1 | 0 | 1 |
| Total sibships | 69 | 46 | 115 | 15 | 12 | 27 | 3 | 0 | 3 | 1 | 0 | 1 |
| Total uncorrected sib pairs* | 69 | 46 | 115 | 45 | 36 | 81 | 18 | 0 | 18 | 10 | 0 | 10 |
| Total corrected sib pairs** | 69 | 46 | 115 | 30 | 24 | 54 | 9 | 0 | 9 | 4 | 0 | 4 |
| Total sib pairs in entire data set: 224 uncorrected, 182 corrected | | | | | | | | | | | | |

*Counting all possible pairs; a sibship of size s will have $s(s-1)/2$ possible pairs.

** Corrected for greater than 2 sibs in a sibship using the formula $2n/s$ where n is number of possible pairs and s is number of affected sibs in a sibship.