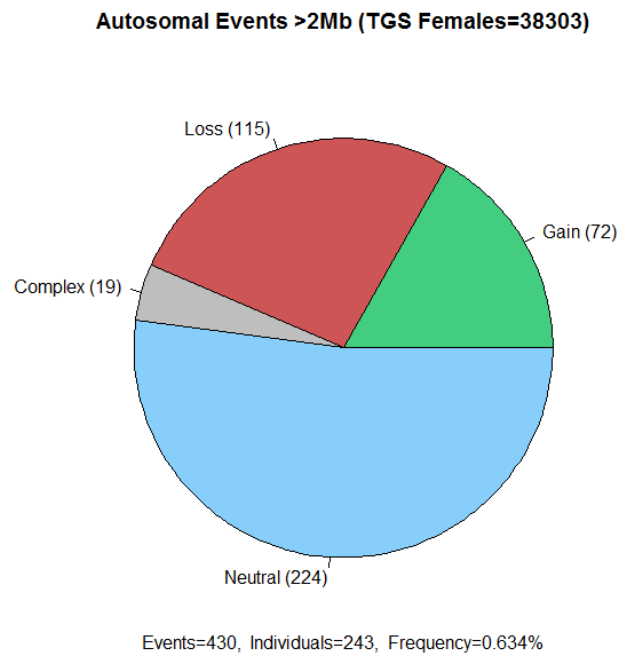
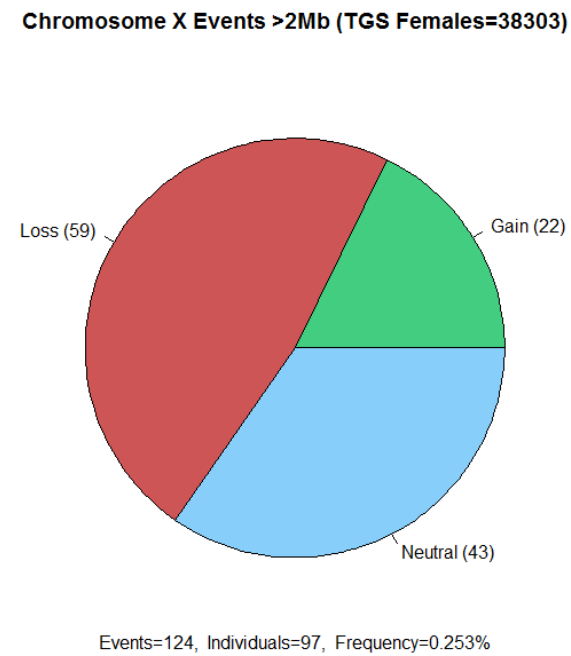


**Supplementary Figure 1. Distribution of detected mosaic events by copy number state for (A) the autosomes and (B) the X chromosome.** Labels indicate event copy number state and numbers in parentheses are counts for total number of detected events.

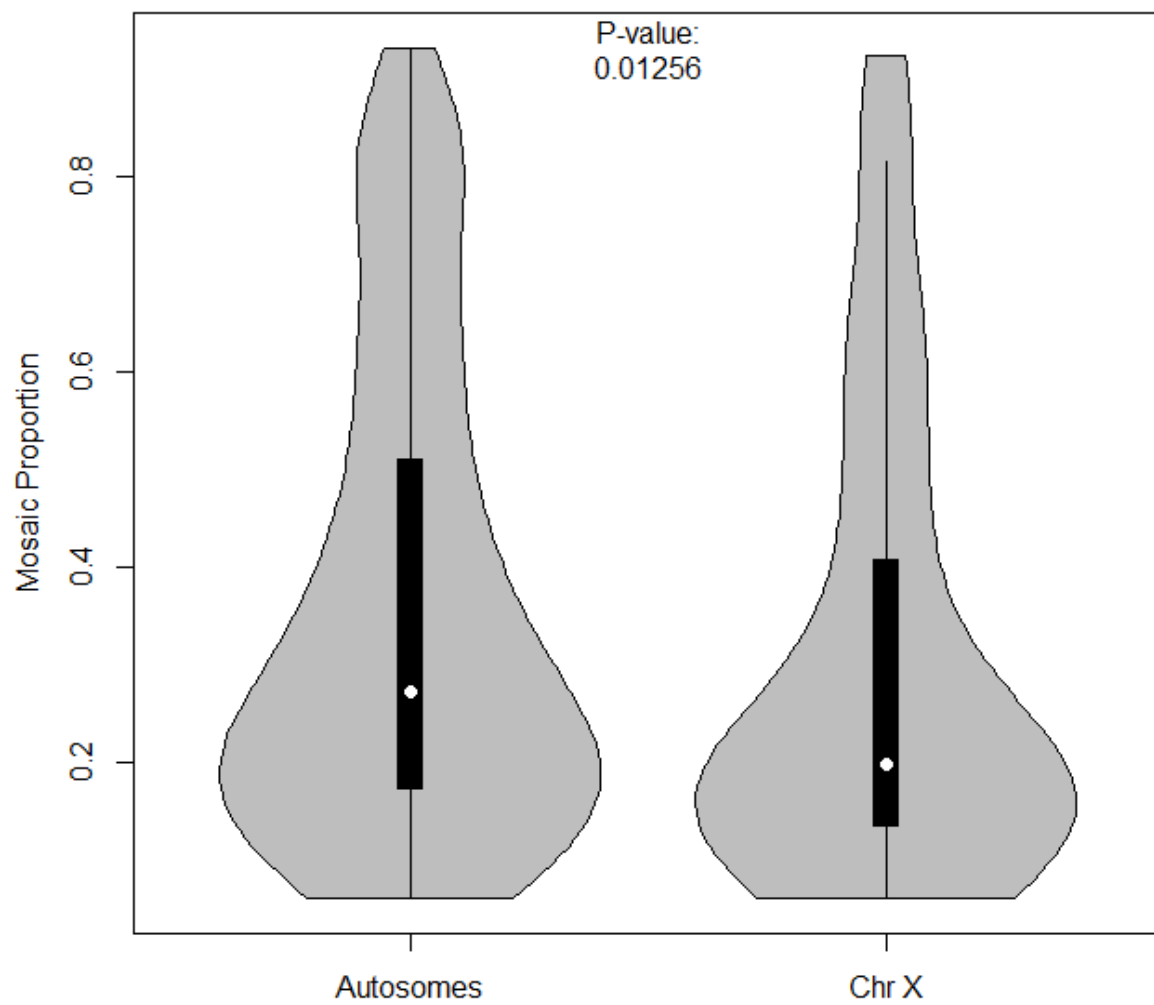
**(A)**



**(B)**

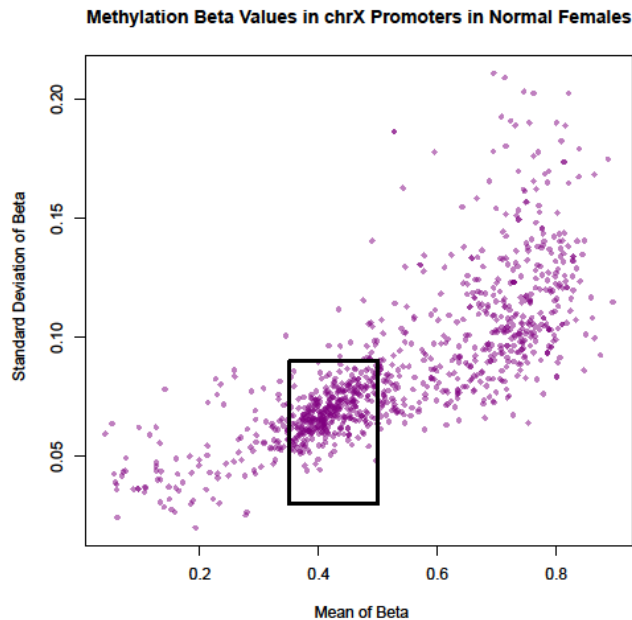


**Supplementary Figure 2. Distribution of mosaic proportions for detected mosaic X events in comparison to detected autosomal mosaic events.** P-value tests for a difference in distributional means.

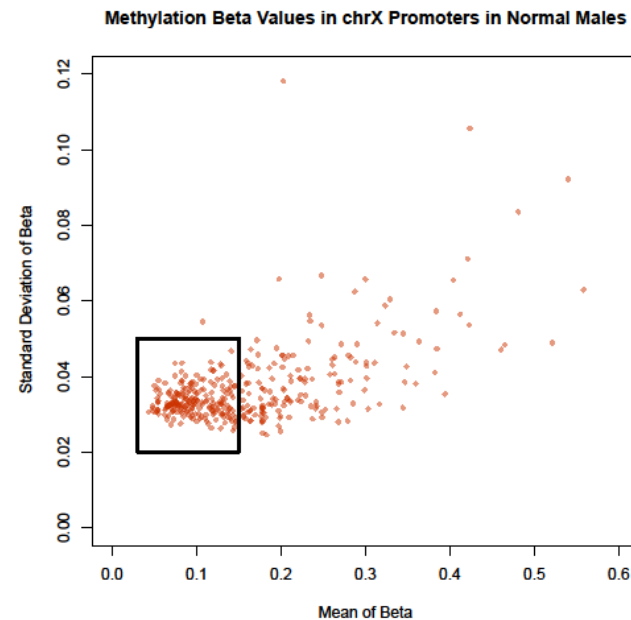


**Supplementary Figure 3. Mean beta values and standard deviations for methylation probe promoter regions for the Illumina Methy450 array in 1,665 control men and 136 control women.** (A) Promoter regions selected in women had mean beta values between 0.35 and 0.5 and standard deviations less than 0.09. (B) The filtered promoter probes in women were investigated in men and further filtered to only include promoter probe regions with mean beta values less than 0.15 and standard deviations less than 0.05. In total, 1,888 differentially methylated probes that spanned 212 promoter sites across the X chromosome were included in subsequent analyses.

(A)



(B)



**Supplementary Table 1. Locations and individual characteristics of detected chromosome X events.**

| ID | Chr | Start       | End         | Size        | Probes | State   | Mosaic Percent | Sex    | Age DNA | Cancer      |
|----|-----|-------------|-------------|-------------|--------|---------|----------------|--------|---------|-------------|
| 1  | X   | 1,553       | 154,582,606 | 154,581,053 | 13,331 | Loss    | 0.511          | Female | NA      | Gastric     |
| 2  | X   | 31,108,125  | 33,712,571  | 2,604,446   | 642    | Neutral | 0.510          | Female | 71      | Stomach     |
| 3  | X   | 132,234,466 | 154,582,606 | 22,348,140  | 2,634  | Neutral | 0.092          | Female | 65      | Cancer-free |
| 4  | X   | 1,553       | 154,582,606 | 154,581,053 | 13,906 | Loss    | 0.689          | Female | NA      | Lung        |
| 5  | X   | 67,840,460  | 154,446,085 | 86,605,625  | 6,921  | Loss    | 0.904          | Female | 39      | Lung        |
| 6  | X   | 38,541,601  | 41,214,219  | 2,672,618   | 346    | Neutral | 0.137          | Female | 61      | Breast      |
| 7  | X   | 45,000,000  | 134,901,714 | 89,901,714  | 20,916 | Gain    | 0.198          | Female | 71      | Cancer-free |
| 8  | X   | 9,120,838   | 20,667,813  | 11,546,975  | 1,345  | Neutral | 0.124          | Female | 54      | Cancer-free |
| 8  | X   | 37,332,880  | 47,772,472  | 10,439,592  | 1,044  | Neutral | 0.134          | Female | 54      | Cancer-free |
| 8  | X   | 117,279,682 | 119,548,573 | 2,268,891   | 231    | Neutral | 0.250          | Female | 54      | Cancer-free |
| 8  | X   | 152,290,349 | 154,553,040 | 2,262,691   | 155    | Neutral | 0.350          | Female | 54      | Cancer-free |
| 9  | X   | 140,204,533 | 146,339,953 | 6,135,420   | 832    | Neutral | 0.118          | Female | 66      | Cancer-free |
| 9  | X   | 22,000,000  | 34,450,748  | 12,450,748  | 1,886  | Neutral | 0.119          | Female | 66      | Cancer-free |
| 9  | X   | 78,809,020  | 99,500,000  | 20,690,980  | 1,429  | Neutral | 0.119          | Female | 66      | Cancer-free |
| 9  | X   | 114,658,108 | 128,000,000 | 13,341,892  | 1,338  | Neutral | 0.119          | Female | 66      | Cancer-free |
| 10 | X   | 1,553       | 154,582,606 | 154,581,053 | 12,246 | Loss    | 0.157          | Female | 63      | Lung        |
| 11 | X   | 111,317,946 | 152,522,337 | 41,204,391  | 4,341  | Loss    | 0.182          | Female | 46      | Lung        |
| 12 | X   | 1,553       | 154,579,620 | 154,578,067 | 13,311 | Loss    | 0.134          | Female | 66      | Breast      |
| 13 | X   | 9,402,279   | 20,000,000  | 10,597,721  | 3,606  | Neutral | 0.172          | Female | 67      | Cancer-free |
| 13 | X   | 37,595,757  | 48,938,568  | 11,342,811  | 3,513  | Neutral | 0.213          | Female | 67      | Cancer-free |
| 14 | X   | 99,148,316  | 129,076,960 | 29,928,644  | 3,502  | Loss    | 0.660          | Female | NA      | Cancer-free |
| 15 | X   | 12,859,359  | 22,057,972  | 9,198,613   | 805    | Neutral | 0.100          | Female | 60      | Breast      |
| 15 | X   | 36,000,000  | 48,000,000  | 12,000,000  | 1,124  | Neutral | 0.100          | Female | 60      | Breast      |
| 16 | X   | 1,553       | 155,260,560 | 155,259,007 | 13,282 | Gain    | 0.825          | Female | 56      | Cancer-free |
| 17 | X   | 9,225,116   | 22,000,000  | 12,774,884  | 1,835  | Neutral | 0.210          | Female | NA      | Endometrial |
| 17 | X   | 35,000,000  | 56,000,000  | 21,000,000  | 2,285  | Gain    | 0.649          | Female | NA      | Endometrial |
| 18 | X   | 1,553       | 155,260,560 | 155,259,007 | 39,903 | Loss    | 0.351          | Female | 50      | Cancer-free |
| 19 | X   | 2,500,000   | 154,582,606 | 152,082,606 | 17,998 | Loss    | 0.383          | Female | NA      | Endometrial |
| 20 | X   | 1,553       | 155,260,560 | 155,259,007 | 38,666 | Loss    | 0.209          | Female | 80      | Cancer-free |
| 21 | X   | 1,553       | 155,260,560 | 155,259,007 | 13,287 | Loss    | 0.168          | Female | 63      | Multiple    |
| 22 | X   | 1,553       | 155,260,560 | 155,259,007 | 13,235 | Gain    | 0.839          | Female | 62      | Glioma      |
| 23 | X   | 1,553       | 154,582,606 | 154,581,053 | 12,787 | Gain    | 0.206          | Female | 52      | Lung        |
| 24 | X   | 38,482,237  | 41,458,510  | 2,976,273   | 374    | Neutral | 0.138          | Female | 77      | Breast      |
| 25 | X   | 1,553       | 154,579,620 | 154,578,067 | 40,102 | Loss    | 0.179          | Female | 77      | Lung        |
| 26 | X   | 37,332,880  | 41,458,510  | 4,125,630   | 514    | Neutral | 0.091          | Female | 73      | Breast      |
| 26 | X   | 15,000,000  | 21,000,000  | 6,000,000   | 499    | Neutral | 0.097          | Female | 73      | Breast      |
| 27 | X   | 3,427,253   | 154,443,832 | 151,016,579 | 13,101 | Loss    | 0.353          | Female | 71      | Cancer-free |
| 28 | X   | 115,322,150 | 154,090,810 | 38,768,660  | 5,681  | Neutral | 0.077          | Female | NA      | Cancer-free |
| 29 | X   | 2,500,000   | 154,582,606 | 152,082,606 | 17,892 | Gain    | 0.277          | Female | NA      | Endometrial |
| 30 | X   | 1,553       | 154,582,606 | 154,581,053 | 13,338 | Loss    | 0.348          | Female | NA      | Bladder     |
| 31 | X   | 2,500,000   | 154,582,606 | 152,082,606 | 18,098 | Loss    | 0.643          | Female | NA      | Cancer-free |

|    |   |             |             |             |        |         |       |        |    |             |
|----|---|-------------|-------------|-------------|--------|---------|-------|--------|----|-------------|
| 32 | X | 2,724,756   | 154,582,606 | 151,857,850 | 14,057 | Gain    | 0.866 | Female | 49 | Lung        |
| 33 | X | 2,735,883   | 154,582,606 | 151,846,723 | 39,632 | Loss    | 0.155 | Female | 67 | Lung        |
| 34 | X | 9,120,838   | 20,473,147  | 11,352,309  | 1,321  | Neutral | 0.158 | Female | 64 | Breast      |
| 34 | X | 37,332,880  | 47,772,472  | 10,439,592  | 1,045  | Neutral | 0.178 | Female | 64 | Breast      |
| 34 | X | 117,279,682 | 119,548,573 | 2,268,891   | 233    | Neutral | 0.178 | Female | 64 | Breast      |
| 35 | X | 1,553       | 154,582,606 | 154,581,053 | 13,324 | Loss    | 0.719 | Female | NA | Unknown     |
| 36 | X | 125,683,733 | 154,138,131 | 28,454,398  | 2,966  | Gain    | 0.245 | Female | 38 | Lung        |
| 37 | X | 1,553       | 154,582,606 | 154,581,053 | 13,336 | Loss    | 0.307 | Female | NA | Unknown     |
| 38 | X | 30,548,244  | 33,300,070  | 2,751,826   | 665    | Loss    | 0.519 | Female | 74 | Esophagus   |
| 39 | X | 27,015,006  | 32,083,817  | 5,068,811   | 766    | Loss    | 0.182 | Female | 56 | Lung        |
| 39 | X | 2,835,403   | 16,000,000  | 6,343,174   | 764    | Gain    | 0.222 | Female | 56 | Lung        |
| 40 | X | 2,500,000   | 154,582,606 | 152,082,606 | 17,860 | Loss    | 0.230 | Female | NA | Endometrial |
| 41 | X | 1,553       | 154,582,606 | 154,581,053 | 12,972 | Loss    | 0.324 | Female | 70 | Lung        |
| 42 | X | 1,553       | 154,582,606 | 154,581,053 | 12,551 | Gain    | 0.880 | Female | 68 | Lung        |
| 43 | X | 143,004,816 | 145,031,290 | 2,026,474   | 564    | Loss    | 0.487 | Female | 70 | Cancer-free |
| 44 | X | 1,553       | 155,260,560 | 155,259,007 | 13,260 | Loss    | 0.168 | Female | 62 | Esophagus   |
| 45 | X | 36,000,000  | 50,000,000  | 14,000,000  | 1,151  | Neutral | 0.095 | Female | 64 | Lung        |
| 45 | X | 111,478,662 | 154,712,424 | 43,233,762  | 4,337  | Neutral | 0.198 | Female | 64 | Lung        |
| 45 | X | 2,787,560   | 15,086,954  | 12,299,394  | 1,554  | Neutral | 0.225 | Female | 64 | Lung        |
| 45 | X | 92,000,000  | 96,000,000  | 4,000,000   | 365    | Gain    | 0.242 | Female | 64 | Lung        |
| 46 | X | 1,553       | 154,582,606 | 154,581,053 | 13,342 | Loss    | 0.663 | Female | NA | Bladder     |
| 47 | X | 1,553       | 154,579,620 | 154,578,067 | 39,220 | Loss    | 0.143 | Female | 68 | Cancer-free |
| 48 | X | 1,553       | 154,582,606 | 154,581,053 | 13,332 | Gain    | 0.157 | Female | NA | Gastric     |
| 49 | X | 1,553       | 155,260,560 | 155,259,007 | 13,280 | Loss    | 0.434 | Female | 77 | Cancer-free |
| 50 | X | 2,519       | 72,267,780  | 72,265,261  | 6,316  | Gain    | 0.129 | Female | 68 | Breast      |
| 50 | X | 72,267,781  | 155,260,560 | 82,992,779  | 6,875  | Loss    | 0.214 | Female | 68 | Breast      |
| 51 | X | 94,926,466  | 127,956,023 | 30,029,557  | 2,457  | Neutral | 0.097 | Female | 60 | Lung        |
| 51 | X | 135,049,024 | 152,850,324 | 17,801,300  | 2,167  | Loss    | 0.148 | Female | 60 | Lung        |
| 52 | X | 148,371,055 | 152,616,777 | 4,245,722   | 560    | Neutral | 0.080 | Female | 70 | Cancer-free |
| 52 | X | 37,332,880  | 48,895,426  | 11,562,546  | 1,093  | Neutral | 0.090 | Female | 70 | Cancer-free |
| 53 | X | 2,500,000   | 154,582,606 | 152,082,606 | 17,821 | Loss    | 0.253 | Female | NA | Endometrial |
| 54 | X | 38,551,737  | 41,483,959  | 2,932,222   | 908    | Neutral | 0.100 | Female | 77 | Lung        |
| 55 | X | 37,537,457  | 48,000,000  | 10,462,543  | 1,479  | Neutral | 0.162 | Female | NA | Cancer-free |
| 56 | X | 85,385,491  | 154,477,684 | 69,092,193  | 19,160 | Neutral | 0.079 | Female | 75 | Cancer-free |
| 57 | X | 21,691,798  | 36,511,216  | 14,819,418  | 2,056  | Neutral | 0.100 | Female | 49 | Lung        |
| 57 | X | 77,009,442  | 98,330,836  | 21,321,394  | 1,449  | Neutral | 0.100 | Female | 49 | Lung        |
| 57 | X | 111,316,727 | 125,023,781 | 13,707,054  | 1,367  | Neutral | 0.100 | Female | 49 | Lung        |
| 57 | X | 140,000,000 | 148,000,000 | 8,000,000   | 1,038  | Neutral | 0.100 | Female | 49 | Lung        |
| 58 | X | 1,553       | 154,582,606 | 154,581,053 | 12,255 | Loss    | 0.126 | Female | 56 | Cancer-free |
| 59 | X | 108,000,000 | 115,904,104 | 7,904,104   | 821    | Loss    | 0.923 | Female | NA | Cancer-free |
| 60 | X | 2,500,000   | 154,582,606 | 152,082,606 | 17,884 | Loss    | 0.887 | Female | NA | Endometrial |
| 61 | X | 1,553       | 155,260,560 | 155,259,007 | 13,072 | Loss    | 0.625 | Female | NA | Cancer-free |
| 62 | X | 1,553       | 155,260,560 | 155,259,007 | 13,288 | Gain    | 0.821 | Female | 54 | Cancer-free |
| 63 | X | 2,500,000   | 154,582,606 | 152,082,606 | 17,830 | Loss    | 0.218 | Female | NA | Cancer-free |

|    |   |             |             |             |        |         |       |        |     |             |
|----|---|-------------|-------------|-------------|--------|---------|-------|--------|-----|-------------|
| 64 | X | 1,553       | 154,579,620 | 154,578,067 | 13,257 | Loss    | 0.136 | Female | 65  | Breast      |
| 65 | X | 12,000,000  | 22,078,734  | 10,078,734  | 3,246  | Neutral | 0.099 | Female | 62  | Cancer-free |
| 65 | X | 128,264,436 | 134,901,714 | 6,637,278   | 1,943  | Neutral | 0.110 | Female | 62  | Cancer-free |
| 65 | X | 38,056,705  | 75,228,249  | 37,171,544  | 8,227  | Neutral | 0.124 | Female | 62  | Cancer-free |
| 65 | X | 99,732,340  | 109,706,724 | 9,974,384   | 3,049  | Neutral | 0.128 | Female | 62  | Cancer-free |
| 65 | X | 117,331,631 | 119,971,453 | 2,639,822   | 826    | Neutral | 0.147 | Female | 62  | Cancer-free |
| 66 | X | 99,573,032  | 114,378,172 | 14,805,140  | 865    | Gain    | 0.862 | Female | 58  | Lung        |
| 67 | X | 1,553       | 154,582,606 | 154,581,053 | 12,783 | Gain    | 0.653 | Female | 67  | Lung        |
| 68 | X | 1,553       | 155,260,560 | 155,259,007 | 14,971 | Loss    | 0.569 | Female | 67  | Lung        |
| 69 | X | 28,343,230  | 31,443,128  | 3,099,898   | 436    | Loss    | 0.620 | Female | NA  | Cancer-free |
| 70 | X | 36,511,216  | 55,616,996  | 19,105,780  | 1,646  | Neutral | 0.095 | Female | 72  | Cancer-free |
| 71 | X | 2,787,560   | 154,582,606 | 151,795,046 | 13,813 | Loss    | 0.185 | Female | 78  | Cancer-free |
| 72 | X | 39,483,015  | 53,636,899  | 14,153,884  | 1,156  | Loss    | 0.182 | Female | 68  | Lung        |
| 73 | X | 1,553       | 155,260,560 | 155,259,007 | 13,736 | Loss    | 0.605 | Female | 44. | Breast      |
| 74 | X | 1,553       | 155,260,560 | 155,259,007 | 13,729 | Loss    | 0.199 | Female | 65  | Breast      |
| 75 | X | 1,553       | 155,260,560 | 155,259,007 | 13,632 | Loss    | 0.369 | Female | 74  | Lung        |
| 76 | X | 2,750,681   | 154,450,686 | 151,700,005 | 14,954 | Loss    | 0.451 | Female | 62  | Cancer-free |
| 77 | X | 1,553       | 155,260,560 | 155,259,007 | 14,976 | Gain    | 0.157 | Female | 60  | Bladder     |
| 78 | X | 1,553       | 155,260,560 | 155,259,007 | 15,012 | Gain    | 0.138 | Female | 80  | Lung        |
| 79 | X | 1,553       | 155,260,560 | 155,259,007 | 14,917 | Loss    | 0.175 | Female | 80  | Lung        |
| 80 | X | 1,553       | 155,260,560 | 155,259,007 | 13,773 | Loss    | 0.142 | Female | 71  | Pancreas    |
| 81 | X | 1,553       | 155,260,560 | 155,259,007 | 14,956 | Loss    | 0.250 | Female | 54  | Pancreas    |
| 82 | X | 1,553       | 155,260,560 | 155,259,007 | 15,044 | Loss    | 0.404 | Female | 79  | Cancer-free |
| 83 | X | 1,553       | 155,260,560 | 155,259,007 | 14,956 | Loss    | 0.268 | Female | 78  | Lung        |
| 84 | X | 1,553       | 155,260,560 | 155,259,007 | 15,041 | Loss    | 0.526 | Female | 72  | Cancer-free |
| 85 | X | 1,553       | 155,260,560 | 155,259,007 | 13,365 | Gain    | 0.638 | Female | 54  | Cancer-free |
| 86 | X | 12,500,000  | 50,000,000  | 37,500,000  | 4,470  | Neutral | 0.062 | Female | 66  | Lung        |
| 87 | X | 33,450,778  | 125,592,155 | 92,141,377  | 7,377  | Loss    | 0.152 | Female | 73  | Cancer-free |
| 87 | X | 125,592,156 | 153,890,083 | 28,297,927  | 3,249  | Gain    | 0.552 | Female | 73  | Cancer-free |
| 88 | X | 1,553       | 155,260,560 | 155,259,007 | 13,363 | Loss    | 0.229 | Female | 66  | Glioma      |
| 89 | X | 1,553       | 155,260,560 | 155,259,007 | 14,963 | Loss    | 0.373 | Female | 82  | Cancer-free |
| 90 | X | 1,553       | 155,260,560 | 155,259,007 | 15,034 | Gain    | 0.193 | Female | 72  | Bladder     |
| 91 | X | 1,553       | 155,260,560 | 155,259,007 | 33,108 | Loss    | 0.469 | Female | 70  | Bladder     |
| 92 | X | 1,553       | 155,260,560 | 155,259,007 | 14,989 | Loss    | 0.208 | Female | 68  | Kidney      |
| 93 | X | 1,553       | 154,579,620 | 154,578,067 | 14,976 | Loss    | 0.150 | Female | 71  | Cancer-free |
| 94 | X | 77,414,128  | 153,886,968 | 76,472,840  | 6,995  | Gain    | 0.213 | Female | 74  | Cancer-free |
| 95 | X | 1,553       | 155,260,560 | 155,259,007 | 14,999 | Loss    | 0.419 | Female | 82  | Lung        |
| 96 | X | 2,750,681   | 154,582,606 | 151,831,925 | 15,019 | Loss    | 0.170 | Female | 64  | Cancer-free |
| 97 | X | 1,553       | 154,579,620 | 154,578,067 | 14,972 | Loss    | 0.140 | Female | 68  | Cancer-free |

**Supplementary Table 2. Mosaic X events by contributing study.**

|                      | No X Mosaicism | X Mosaicism | Total |
|----------------------|----------------|-------------|-------|
| AHS                  | 20             | 1           | 21    |
| AustraliaCancerStudy | 29             | 0           | 29    |
| Beijing              | 791            | 0           | 791   |
| Brazil               | 51             | 0           | 51    |
| CeRePP               | 38             | 1           | 39    |
| CLUEII               | 74             | 1           | 75    |
| CNULCS               | 1,080          | 4           | 1,084 |
| COG                  | 265            | 0           | 265   |
| CONN                 | 1,031          | 2           | 1,033 |
| CONN-OLD             | 66             | 0           | 66    |
| CPSII                | 1,618          | 13          | 1,631 |
| EAGLE                | 860            | 1           | 861   |
| EPIC                 | 801            | 2           | 803   |
| FBCS                 | 30             | 1           | 31    |
| FHCRC                | 1,401          | 3           | 1,404 |
| FTCS                 | 69             | 0           | 69    |
| Fudan                | 584            | 0           | 584   |
| GELAC                | 1,198          | 3           | 1,201 |
| Gliogene             | 145            | 1           | 146   |
| GROUPHEALTH          | 272            | 1           | 273   |
| Guangdong            | 711            | 1           | 712   |
| HERPACC              | 633            | 0           | 633   |
| HKLymphoma           | 378            | 1           | 379   |
| HPFS                 | 1              | 1           | 2     |
| Interphone           | 269            | 0           | 269   |
| Italy                | 21             | 0           | 21    |
| JHU                  | 235            | 0           | 235   |
| Korea-Univ           | 103            | 0           | 103   |
| Kyungpook-Univ       | 221            | 1           | 222   |
| MAYO                 | 579            | 0           | 579   |
| MCCS                 | 54             | 0           | 54    |
| MDA                  | 222            | 0           | 222   |
| MDA_Amos_Spitz_Wu    | 354            | 1           | 355   |
| MDA_McNeil           | 595            | 0           | 595   |
| MEC                  | 641            | 1           | 642   |
| Melbourne            | 23             | 0           | 23    |
| MSKCC                | 149            | 0           | 149   |
| NCC                  | 278            | 0           | 278   |
| NCI_BTS              | 366            | 1           | 367   |
| NCI-UMD              | 277            | 2           | 279   |
| NEBCS                | 296            | 1           | 297   |

|               |               |           |               |
|---------------|---------------|-----------|---------------|
| NEET          | 593           | 3         | 596           |
| NHS           | 3,397         | 3         | 3,400         |
| NHS2          | 546           | 1         | 547           |
| NIOSH_AGCC    | 357           | 1         | 358           |
| NITC          | 606           | 3         | 609           |
| NSHDS         | 477           | 0         | 477           |
| NYU-WHS       | 33            | 0         | 33            |
| Osteosarcoma  | 37            | 0         | 37            |
| PamplonaCUN   | 37            | 0         | 37            |
| PBCS          | 1,140         | 7         | 1,147         |
| PECC          | 487           | 0         | 487           |
| PeterMac      | 3             | 0         | 3             |
| PLCO          | 2,804         | 13        | 2,817         |
| POCC          | 260           | 0         | 260           |
| Poland        | 155           | 0         | 155           |
| SBCS          | 261           | 1         | 262           |
| SCCS          | 203           | 1         | 204           |
| Shenyang      | 1,007         | 5         | 1,012         |
| SHNX          | 1,134         | 0         | 1,134         |
| SING          | 129           | 0         | 129           |
| SINGAPORE     | 493           | 0         | 493           |
| SMWHS         | 124           | 0         | 124           |
| SNU           | 162           | 0         | 162           |
| SNUH          | 44            | 0         | 44            |
| SNUPM         | 83            | 0         | 83            |
| Sweden        | 31            | 0         | 31            |
| SWHS          | 619           | 0         | 619           |
| Taiwan        | 1,031         | 1         | 1,032         |
| Tianjin       | 478           | 1         | 479           |
| TORONTO       | 260           | 1         | 261           |
| UCL_UK        | 15            | 0         | 15            |
| UCSF          | 750           | 3         | 753           |
| USMulticenter | 8             | 0         | 8             |
| USRC          | 773           | 2         | 775           |
| WayneState    | 355           | 1         | 356           |
| WHI           | 976           | 3         | 979           |
| WHS           | 57            | 0         | 57            |
| Wuhan         | 63            | 0         | 63            |
| Xuan-Wei      | 1,155         | 2         | 1,157         |
| YALE          | 234           | 1         | 235           |
| <b>Total</b>  | <b>38,206</b> | <b>97</b> | <b>38,303</b> |



**Supplementary Table 3. Chromosomal arm location by copy number state.**

|                  | <b>Gain</b> |       | <b>Loss</b> |       | <b>Neutral</b> |       | <b>Total</b> |      |
|------------------|-------------|-------|-------------|-------|----------------|-------|--------------|------|
| interstitial     | 3           | 6.5%  | 8           | 17.4% | 35             | 76.1% | 46           | 100% |
| spans centromere | 1           | 50.0% | 0           | 0.0%  | 1              | 50.0% | 2            | 100% |
| telomeric p      | 2           | 50.0% | 0           | 0.0%  | 2              | 50.0% | 4            | 100% |
| telomeric q      | 3           | 25.0% | 4           | 33.3% | 5              | 41.7% | 12           | 100% |
| whole            | 13          | 21.7% | 47          | 78.3% | 0              | 0.0%  | 60           | 100% |
|                  | 22          | 17.7% | 59          | 47.6% | 43             | 34.7% | 124          | 100% |

**Supplementary Table 4. Validation qPCR probes and locations.**

| SNP or<br>GENE<br>Symbol | Gene Name                                            | Chr | Location<br>(NCBI Build 37) | Life Assay ID | NCBI Location            | Cytoband<br>Location | Assay Location               | Amplicon<br>Length |
|--------------------------|------------------------------------------------------|-----|-----------------------------|---------------|--------------------------|----------------------|------------------------------|--------------------|
| XG                       | Xg blood group                                       | X   | 2703859 Hs04100849_cn       |               | ChrX:2703633-2805662     | Xp22.33c             | Within Intron 4              | 82                 |
| PHEX                     | phosphate regulating endopeptidase homolog; X-linked | X   | 22053233 Hs05679343_cn      |               | ChrX:22050921-22266478   | Xp22.11b             | Within Intron 1              | 86                 |
| CXorf22                  | chromosome X open reading frame 22                   | X   | 35937936 Hs01859438_cn      |               | ChrX:35937851-36008269   | Xp21.1b              | Within Exon 1                | 88                 |
| KDM6A                    | lysine (K)-specific demethylase 6A                   | X   | 44736584 Hs05632292_cn      |               | ChrX:44732423-44971847   | Xp11.3b              | Within Intron 2              | 98                 |
| CXorf65                  | chromosome X open reading frame 65                   | X   | 70325895 Hs00598842_cn      |               | ChrX:69640535-71108033   | Xq13.1d              | Within Exon 3                | 80                 |
| PCDH11X                  | protocadherin 11 X-linked                            | X   | 91127540 Hs05604144_cn      |               | ChrX:91099385-91199647   | Xq21.31e             | Within Intron 4              | 104                |
| CXorf57                  | chromosome X open reading frame 57                   | X   | 105922389 Hs02805049_cn     |               | ChrX:104949928-106367102 | Xq22.3b              | Within Exon 14               | 110                |
| CXorf61                  | chromosome X open reading frame 61                   | X   | 115593024 Hs01583891_cn     |               | ChrX:115589779-115624958 | Xq23e                | Within Exon 2                | 101                |
| CXorf64                  | chromosome X open reading frame 64                   | X   | 125954155 Hs04097898_cn     |               | ChrX:125930015-125965957 | Xq25f                | Within Intron 1              | 94                 |
| RBMX                     | RNA binding motif protein; X-linked                  | X   | 135952571 Hs05654346_cn     |               | ChrX:135951353-135962939 | Xq26.3c              | Overlaps Intron 10 - Exon 11 | 107                |
| SPANXN3                  | SPANX family; member N3                              | X   | 142596950 Hs02664978_cn     |               | ChrX:142594742-142798873 | Xq27.3a              | Overlaps Intron 1 - Exon 2   | 109                |
| PLXNA3                   | plexin A3                                            | X   | 153701896 Hs01014974_cn     |               | ChrX:153348431-153829693 | Xq28g                | Within Exon 33               | 103                |
| NLGN4Y                   | neuroligin 4; Y-linked                               | Y   | 16680289 Hs05707486_cn      |               | ChrY:16634488-16955848   | Yq11.221c            | Within Intron 4              | 104                |

**Supplementary Table 5. Validation results from qPCR and concordance rates with array genotyping.**

|               |         | Whole Chromosome<br>qPCR   |         |      |        |    | <u>Concordance</u> |
|---------------|---------|----------------------------|---------|------|--------|----|--------------------|
|               |         | Loss                       | Neutral | Gain | Failed |    |                    |
| GWAS<br>Array | Loss    | 12                         | 2       | 1    | 0      | 15 | 80.00%             |
|               | Neutral | 0                          | 0       | 0    | 0      | 0  | NA                 |
|               | Gain    | 0                          | 0       | 3    | 0      | 3  | 100.00%            |
|               |         | 12                         | 2       | 4    | 0      | 18 | 83.33%             |
|               |         | Partial Chromosome<br>qPCR |         |      |        |    | <u>Concordance</u> |
|               |         | Loss                       | Neutral | Gain | Failed |    |                    |
| GWAS<br>Array | Loss    | 0                          | 1       | 1    | 0      | 2  | 0.00%              |
|               | Neutral | 0                          | 1       | 2    | 2      | 5  | 20.00%             |
|               | Gain    | 0                          | 0       | 1    | 0      | 1  | 100.00%            |
|               |         | 0                          | 2       | 4    | 2      | 8  | 25.00%             |

**Supplementary Table 6. Average beta and percent mosaic values for methylation samples.**

| <b>Sample</b> | <b>Copy Number State<sup>1</sup></b> | <b>Average Beta<sup>2</sup></b> | <b>Percent Mosaic<sup>1</sup></b> |
|---------------|--------------------------------------|---------------------------------|-----------------------------------|
| 1             | Loss                                 | 0.280                           | 0.134                             |
| 2             | Loss                                 | 0.303                           | 0.569                             |
| 3             | Loss                                 | 0.323                           | 0.605                             |
| 4             | Loss                                 | 0.331                           | 0.229                             |
| 5             | Loss                                 | 0.339                           | 0.469                             |
| 6             | Loss                                 | 0.343                           | 0.208                             |
| 7             | Loss                                 | 0.354                           | 0.404                             |
| 8             | Loss                                 | 0.368                           | 0.143                             |
| 9             | Loss                                 | 0.370                           | 0.199                             |
| 10            | Loss                                 | 0.380                           | 0.157                             |
| 11            | Loss                                 | 0.387                           | 0.168                             |
| 12            | Loss                                 | 0.389                           | 0.142                             |
| 13            | Neutral                              | 0.389                           | 0.100                             |
| 14            | Loss                                 | 0.392                           | 0.152                             |
| 15            | Loss                                 | 0.398                           | 0.140                             |
| 16            | Loss                                 | 0.406                           | 0.136                             |
| 17            | Loss                                 | 0.408                           | 0.268                             |
| 18            | Loss                                 | 0.422                           | 0.519                             |
| 19            | Loss                                 | 0.425                           | 0.150                             |
| 20            | Loss                                 | 0.437                           | 0.175                             |
| 21            | Neutral                              | 0.448                           | 0.091                             |
| 22            | Loss                                 | 0.452                           | 0.526                             |
| 23            | Neutral                              | 0.455                           | 0.062                             |
| 24            | Gain                                 | 0.464                           | 0.825                             |
| 25            | Neutral                              | 0.469                           | 0.158                             |
| 26            | Loss                                 | 0.483                           | 0.419                             |
| 27            | Neutral                              | 0.486                           | 0.118                             |
| 28            | Neutral                              | 0.501                           | 0.138                             |
| 29            | Neutral                              | 0.507                           | 0.100                             |
| 30            | Neutral                              | 0.510                           | 0.100                             |
| 31            | Gain                                 | 0.510                           | 0.157                             |
| 32            | Gain                                 | 0.513                           | 0.193                             |
| 33            | Loss                                 | 0.521                           | 0.182                             |
| 34            | Gain                                 | 0.522                           | 0.138                             |
| 35            | Neutral                              | 0.562                           | 0.172                             |
| 36            | Gain                                 | 0.582                           | 0.198                             |
| 37            | Neutral                              | 0.613                           | 0.137                             |
| 38            | Neutral                              | 0.643                           | 0.210                             |

(1) Copy number state and percent mosaicism determined from SNP genotyping arrays

(2) Beta value from Illumina HumanMethylation450 arrays