

SUPPLEMENTARY INFORMATION

Independent specialization of the human and mouse X chromosomes for the male germline

Jacob L. Mueller¹, Helen Skaletsky^{1,2}, Laura G. Brown^{1,2}, Sara Zaghul¹, Susan Rock⁴, Tina Graves⁴, Katherine Auger⁵, Wesley C. Warren⁴, Richard K. Wilson⁴, David C. Page^{1,2,3}

¹Whitehead Institute, Cambridge, Massachusetts, USA

²Howard Hughes Medical Institute, Massachusetts Institute of Technology, Cambridge, Massachusetts, USA.

³Department of Biology, Massachusetts Institute of Technology, Cambridge, Massachusetts, USA.

⁴The Genome Institute, Washington University School of Medicine, St. Louis, Missouri, USA.

⁵The Wellcome Trust Sanger Institute, Wellcome Trust Genome Campus, Hinxton, Cambridge, United Kingdom.

Table of Contents

Supplementary Note

A. Problems with multi-haplotype assemblies of ampliconic regions

B. Limitations of using whole genome shotgun sequence to infer the evolutionary history of a gene

Supplementary Figures

Supplementary Figure 1. Triangular dot-plots of a revised human X-ampliconic region containing the CT45 gene family

Supplementary Figure 2. Triangular dot-plots of a revised human X-ampliconic region containing the PNMA6 gene family

Supplementary Figure 3. Triangular dot-plots, based on SHIMS assemblies, of newly identified palindromic amplicons

Supplementary Figure 4. Percentage of genes expressed predominantly in the ovary

Supplementary Figure 5. Dot plots of human X chromosome versus six different mammalian X chromosomes

Supplementary Figure 6. Hybrid male-sterility loci in mice that map to the X chromosome

Supplementary Tables

Supplementary Table 1. Reassembling the human X chromosome: 33 regions scrutinized, 29 of which we sequenced using the SHIMS approach

Supplementary Table 2. Genes shared between human and mouse X chromosomes

Supplementary Table 3. Human X-linked genes with no detectable ortholog on the mouse X chromosome

Supplementary Table 4. Mouse X-linked genes with no detectable ortholog on the human X chromosome

Supplementary Table 5. Tallies of X-linked gene classifications depicted graphically in Figure 3

Supplementary Table 6. Tissue expression of X-linked genes shared between human and mouse

Supplementary Table 7. Expression patterns of independently acquired human X-linked genes

Supplementary Table 8. Expression patterns of independently acquired mouse X-linked genes

Supplementary Table 9. Documenting expression of individual members of independently acquired X-linked multicopy and ampliconic gene families in human and mouse

Supplementary Table 10. Expression patterns of all human autosomal genes

Supplementary Table 11. Expression patterns of all mouse autosomal genes

Supplementary Table 12. All OMIM X-linked phenotypes where the molecular basis is known

Supplementary Table 13. Expression patterns between the sexes, in the heart and kidney, of human genes that follow Ohno's law

Supplementary Table 14. Expression patterns between the sexes, in the heart and kidney, of mouse genes that follow Ohno's law