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Uniparental disomy in ES cells

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Homozygous mutant cells can be generated from embryonic stem (ES) cells with a single insertion of a drug-resistance marker by increasing the concentration of the selection drug. In the March Nature Genetics, Lefebvre *et al*. report analysis of the mechanism governing this loss of heterozygosity (LOH) (*Nature Genetics* 2001, **27:**257-258). They used an ES cell line resulting from a cross between two different inbred mouse 129 substrains which could be distinguished by single sequence-length polymorphisms (SSLP). Lefebvre *et al*. studied six different nemomycin-resistance (*neo*) gene insertions following selection with high doses of the drug G418. They found that in all cases homozygous cells exhibited extensive LOH, even at markers 16-66 cM from the *neo* insertion site. The mechanism of LOH therefore appears to involve chromosome loss and duplication, generating regions of uniparental disomy(UDP). Such UDP may affect the expression of imprinted genes on the duplicated chromosome. These observations may complicate functional analysis, but can also be exploited to screen for recessive phenotypes.

References

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2. *Nature Genetics*, [http://genetics.nature.com]

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4. Genealogies of mouse inbred strains.

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