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Conditions for extinction of some lethal alleles of X-linked genes

Some lethal alleles of certain genes can cause the death of the organisms that carry them. Some of these alleles, as could be that responsible of hemophilia, correspond to genes linked to sex chromosomes, especially to X chromosome. If these alleles are dominant, all the carriers die so they are rarely detected due to their rapid elimination from populations. However, recessive lethal alleles only cause death of carrier males and homozygous carrier females, though the last ones must be daughters of a carrier male, so they rarely exist. Heterozygous carrier females are able to live and reproduce. They do not phenotypically express the genetic condition but can pass the lethal allele onto offspring.

In this work, we introduce a multitype bisexual branching process for describing the evolution of the number of individuals carrying the alleles, R and r , of a gene linked to X chromosome. The R allele is considered dominant and the r allele is assumed to be recessive and lethal. Females can have two genotypes: homozygous, RR , and heterozygous, Rr , whereas only R males are able to live. Homozygous and heterozygous females have identical phenotypes so males do not know the genotype of their mates, it can be said that they made a “blind” choice among the two genotypes.

In such a model, we take into account that the offspring of a couple with a homozygous female do not carry the lethal allele. However, couples with heterozygous females can give birth to RR and Rr females and R and r males. Since r males die, Mendelian inheritance ratios of these couples are altered. The total offspring of each couple is modeled through a random variable whose probability distribution is supposed to be different for homozygous and heterozygous females.

We use this model to study the extinction probability of one of these lethal alleles, i.e. under which conditions it will eventually disappear and when it will survive along the generations. Such conditions are expressed in terms of the parameters of the model. In case of non extinction, we investigate the evolution of the number of carriers of such alleles.

Acknowledgements: Research supported by the Ministerio de Ciencia e Innovación, the Junta de Extremadura and the FEDER, grants MTM2009-13248 and GR10118.