Dr Sibert and co-workers comment:

Bucher and Ionasescu are correct in stating that if only isolated cases of Duchenne muscular dystrophy are considered, more than one-third would be expected to be new mutations according to the Haldane rule. However, this in no way affects the conclusions from our data which are: (1) that the creatine phosphokinase distribution of mothers of isolated cases is incompatible with the great majority being carriers, (2) that the distribution is compatible with one-third being new mutations.

Numbers are insufficient to determine the precise proportion of new mutations in this group, but we remain unconvinced that there is any substantial evidence

favouring inequality of mutation rate between the sexes or any other reason for deviation from the Haldane rule in Duchenne muscular dystrophy. We also stress that in genetic counselling of each new case without family history, a prior risk of one-third of such a case representing a new mutation is the correct figure based on the Haldane rule, and in our opinion this is the prior risk that should be used until more definitive tests of carrier status are available.

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