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EDITORIAL

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Is the amelogenin sex test valid?

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In this issue of the International Journal of Legal Medicine there are two contributions on the same topic. Although this is an unusual situation, this has been done deliberately because of the nature of the topic which should be of special interest to the practicing forensic scientist. The point discussed in these two papers is whether the amelogenin sex test is reliable enough for general use in forensic casework. The problem in question has not gone unnoticed in the past. Although Santos et al. (1998) in their publication in Nature Genetics (Reliability of DNA-based sex tests) recommended that another Y-locus (i.e. *SRY*) should routinely be included in any sex test to avoid mis-typing, the message obviously did not come across forcibly enough and there was little response from the forensic community, perhaps because the deletion of the *AMELY* gene reported was only found in 2 unrelated males from Sri Lanka, albeit from a sample size of only 24!

The two publications included in this issue reinforce the argument for additional testing as although still rare, the deletion of the *AMELY* gene has now been reported in 6 out of 29,432 male individuals included in the Austrian database (Steinlechner et al. 2002 Rare failures in the amelogenin sex test) which contains only individuals of European origin. The second paper (Thangaraj et al. 2002 Is the amelogenin gene reliable for gender identification in forensic casework and prenatal diagnosis?) reports an additional 5 out of 270 randomly selected Indian males which could suggest that this phenomenon occurs with a higher frequency (1.85% compared to 0.02%) in the Indian sub-continent. All individuals with the *AMELY* gene deletion reported so far have a normal male appearance and there are obviously no indications of an abnormal visible male phenotype.

Although this is still rare in the population tested in Austria, it is not known (because it has not been tested) whether other populations will also show an increased occurrence in the same way as the Indian or Sri Lankan population samples or if there is some as yet unknown link between the Austrian males and therefore a reason for suspecting sub-population differences. The implications for the forensic community are potentially serious and should this time be considered more professionally than has been done in the past. The incorrect classification of a male as genetically being a "female" has not only the obvious social consequences for the individual involved. The forensic use of the *AMELY* gene in screening exhibits in a rape or murder case for the presence of small amounts of male DNA could mean that none is detected although actually being present and therefore that forensic evidence critical for the apprehension of a rapist or murderer would be overlooked or indeed destroyed. The chance of this happening is at present unknown because of the lack of information on how common this phenomenon actually is in the general population or in sub-populations. Until this question is properly addressed it would be much more appropriate if, as has always been the maxim of forensic science, adequate control measures in the form of additional Y-chromosome marker(s) were to be included in all sex tests performed for detecting the presence of male DNA.

Perhaps the commercial companies producing multiplex kits for the forensic (and other) markets could be persuaded to include an additional safety measure in the form of a further Y marker for confirmation of the sex status of individuals tested.

The Editor

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