Somatic Mutation Study of Hair Roots in an Individual

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INTRODUCTION

Human hair development is a rapid process. While in the growth stage, no other tissue except bone marrow has as high a rate of mitotic activity as the hair root bulb (1). One group of follicle progenitor stem cells derived from the ectoderm can give rise to up to 3 follicle buds (Figure 1). At 16–20 weeks the follicles begin producing hairs. Follicular material clinging to a forcibly pulled hair is usually of ectodermal origin. The head hair growth phase occurs over 3–5 years, and the rest phase, approximately 3 months. The pubic hair growth phase occurs over 4–7 months, and the rest phase lasts from 6–9 months (1). Therefore, a forcibly pulled head hair is likely in growth phase, while a forcibly pulled pubic hair is likely in rest phase.

A high rate of mitosis can increase the opportunity for mutations to occur. A somatic mutation is any permanent change in the sequence of genomic DNA in a somatic cell rather than in the germline. A mosaic is an individual or tissue with at least two cell lines differing in genotype or karyotype derived from a single zygote, while a chimera is an individual composed of cells derived from two genetically different zygotes (2). The data presented in this study provides evidence for mosaicism in multiple pubic hair roots and a single head hair root from a single individual.

HAIR ROOT SAMPLES

Hairs (head, 143; pubic, 47) were collected from one donor by forceful pulling, and 0.5–0.7cm pieces were cut from the root end for DNA extraction. Samples were processed as described for the Tissue and Hair Extraction Kit (3), except that a modified proteinase K digestion buffer with a high concentration of DTT was used. A Biomek®2000 Automation Workstation (Beckman Coulter) carried out the remainder of the extraction as previously described (4).

The AluQuant®Human DNA Quantitation System (f,g) was used as recommended by the manufacturer (5). DNA samples were amplified using the PowerPlex®16 BIO System (b–e). Reduced-volume reactions (12.5µl) were amplified for 31 rather than 32 PCR cycles (6,7). Manufacturer's recommendations were followed for the monoplex reactions (8). Amplified samples were separated on a 3% NuSieve®agarose product gel prior to polyacrylamide gel electrophoresis (PAGE) to estimate PAGE sample loading. PAGE was performed using a 6% PAGE-PLUS™ (Amresco) polyacrylamide gel run for 2 hours at 60W. PAGE gels were analyzed using the FMBIO®II Fluorescence Imaging System (MiraiBio) with the FMBIO®Analysis and STaRCaLL™ software programs.

RESULTS AND DISCUSSION

A triallelic pattern at D18S51 was identified and confirmed by re-amplification of a pulled head hair root sample with the PowerPlex®16 BIO System (Figure 2, Panel A). An aberrant 28,30 D21S11 genotype from a pulled pubic hair root sample was also identified (data not shown; normal genotype 29,30). An aberrant 11,12 genotype at CSF1PO for four pulled pubic hairs was confirmed by re-amplification with the PowerPlex®16 BIO System (Figure 2, Panel B) and by monoplex amplification of two of four samples with DNA extract remaining (Figure 3). The primers for the PowerPlex®16 BIO System and those for the GenePrint®Fluorescent Monoplex STR System, CSF1PO

Figure 1. Hair follicle development. Hair follicle development during embryogenesis and fully formed containing a growing hair (1).
Two nonoverlapping, independent somatic mutations were observed: one at D18S51, one at D21S11 and four at CSF1PO. Rates were estimated by dividing the number of mutations by the total number of hairs typed. 

<table>
<thead>
<tr>
<th>Sample Type</th>
<th>Total Attempted</th>
<th>PCR Product</th>
<th>Full Profile</th>
<th>Number of Mutations</th>
<th>Occurrence Rate</th>
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<tbody>
<tr>
<td>Head Hairs Total</td>
<td>169</td>
<td>147</td>
<td>143</td>
<td>1</td>
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<tr>
<td>Hair Study 3</td>
<td>115</td>
<td>94</td>
<td>90</td>
<td>0</td>
<td>0.006993007 (D18S51 locus)</td>
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<td>24</td>
<td>24</td>
<td>0</td>
<td></td>
</tr>
<tr>
<td>Hair Study 1</td>
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<td>23</td>
<td>23</td>
<td>0</td>
<td></td>
</tr>
<tr>
<td>Repeat Hair Study</td>
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<td>6</td>
<td>6</td>
<td>1</td>
<td></td>
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<tr>
<td>Pubic Hairs Total</td>
<td>71</td>
<td>54</td>
<td>47</td>
<td>5</td>
<td>0.085106383 (CSF1PO locus)</td>
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<tr>
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<td>45</td>
<td>41</td>
<td>5</td>
<td></td>
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<tr>
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<td>3</td>
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**CONCLUSION**

Somatic mutation in the allelic repeat section is the most likely explanation for the anomalous patterns observed at D21S11, D18S51 and CSF1PO. Two nonoverlapping, independent primer sets for the CSF1PO locus produced the same genotyping result, making primer point mutation(s) unlikely at that locus. Similarly, primer point mutations are unlikely at D21S11 and D18S51 since they do not explain the aberrant patterns observed at those loci. Somatic mutations have a low frequency of occurrence, even in hair roots, which have a high mitotic rate. The individual studied is a somatic mosaic because multiple DNA profiles were present within the body, with the findings inconsistent with a chimera. Awareness of somatic mutation is important for forensic biology casework, since somatic mutation in hair roots has been reported (11). It may be informative to record aberrant patterns in casework to help estimate the frequencies of somatic mutations.

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**REFERENCES**