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# Germline mutations at 15 STR loci in the Chinese population

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**Abstract.** Germline mutation of 15 short tandem repeat (STR) loci was studied for over 52,000 parent–offspring allelic transfers in the Chinese population. Mutation rates varied from $<0.056\%$ to 0.507% (FGA, paternal) and the overall paternal to maternal mutation ratio approximately 5% to 1.94% of the mutations could be explained by losses or gains of one repeat unit and there was no evidence for selection between insertion or deletion changes. Possible silent alleles were detected in three of the paternal exclusions. Mutation data accumulated for the Chinese population was compared with those of the American and European populations. 

*Keywords:* Short tandem repeat (STR); Mutation; Chinese

## 1. Introduction

DNA markers currently used for forensic investigation are all highly polymorphic loci with high discrimination power. On the other hand, they are not controlled by selection pressure and are hence more susceptible to mutation [1]. Germline mutations are particularly critical for parentage investigation because links made between a child and the alleged parent could result in a false exclusion. With the rapid increase in the application of PCR short tandem repeat (STR) technology for parentage analysis and mass disaster identification, the study and understanding of germline mutations at STR loci has become increasingly important. We report an analysis of mutations in the Chinese population basing on data obtained from over 1800 cases of parentage testing.

## 2. Materials and methods

Blood samples and buccal swabs were extracted for their DNA using either Chelex [2] or the QIAamp® DNA Blood Mini Kit. The extracted DNA was quantified with either the QuantiBlot® Human DNA Quantitation Kit or the PicoGreen® dsDNA Quantitation Kit.

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PCR amplification was performed using the GenePrint®PowerPlex™16 Kit and subsequent genotyping was performed with either the ABI Prism®377 or ABI Prism®310 sequencers and the automatic fragment sizing software provided.

We assumed a mutation when there was an isolated non-Mendelian inheritance parent(s)/child mismatch provided the likelihood of parentage exceeded 10,000 to 1, not taking the isolated mismatch into consideration. Also, in accordance with literature [3], we have assigned the 'new' allele to the shortest mutational step. If both parents exhibited the same change in allelic size, the origin was classified uncertain.

## 3. Results and discussion

In a total of 52,377 parent/child allele transfers we observed 67 mutations at 14 of the 15 STR loci of the PowerPlex™16 system. All allele transfers at only the THO1 locus were consistent with Mendelian inheritance. Table 1 summarizes the mutations observed. The overall paternal to maternal mutation ratio is approximately 5 to 1 which is close to that estimated for an American population [3] and a Caucasian population in Europe [4]. Nine mutations were classified as uncertain.

Consistent with previous findings [4,5], most of the mutations (63 out of 67, 94%) could be explained by losses or gains of one repeat unit. This observation would support the strict stepwise replication slippage mutation model [6]. Of the remaining four cases, three involved two-step changes and the fourth, a possible three-step change. Our data does not seem to suggest selection between insertion or deletion changes overall, as observed by Sajantila et al. [7]. Interestingly, for the possible three-step change, the phenotypes of the family trio suggest the possible presence of a silent allele. Silent alleles could also have explained for the non-Mendelian inheritance observed in two other cases.

Table 1
Characteristics of observed mutations in the Chinese population

| Locus | Paternal | | | Maternal | | |
|---|---|---|---|---|---|---|
| | No. of meiosis | No. of mutations | Mutation rate (%) | No. of meiosis | No. of mutations | Mutation rate (%) |
| D3S1358 | 1787 | 0 | <0.056 | 1745 | 0 | <0.057 |
| THO1 | 1795 | 0 | <0.056 | 1765 | 0 | <0.057 |
| D21S11 | 1762 | 2 | 0.114 | 1715 | 1 | 0.058 |
| D18S51 | 1757 | 6 | 0.341 | 1710 | 3 | 0.175 |
| Penta E | 1798 | 4 | 0.222 | 1763 | 3 | 0.170 |
| D5S818 | 1794 | 1 | 0.056 | 1764 | 0 | <0.057 |
| D13S317 | 1798 | 2 | 0.111 | 1769 | 0 | <0.057 |
| D7S820 | 1798 | 2 | 0.111 | 1766 | 1 | <0.057 |
| D16S539 | 1803 | 2 | 0.111 | 1773 | 0 | <0.056 |
| CSF1PO | 1795 | 2 | 0.111 | 1767 | 0 | <0.057 |
| Penta D | 1800 | 3 | 0.167 | 1770 | 0 | <0.056 |
| vWA | 1639 | 8 | 0.488 | 1525 | 0 | <0.066 |
| D8S1179 | 1795 | 7 | 0.390 | 1763 | 1 | 0.057 |
| TPOX | 1794 | 1 | 0.056 | 1756 | 0 | <0.057 |
| FGA | 1578 | 8 | 0.507 | 1533 | 1 | 0.065 |
| Total | 26,493 | 48 | | 25,884 | 10 | |

Table 2
Comparison of mutation rates from Chinese and Western populations

| Locus | Paternal mutation rate (%) | | Maternal mutation rate (%) | |
|---|---|---|---|---|
| | AABB | Chinese | AABB | Chinese |
| D3S1358 | 0.131 | <0.056 | 0.015 | <0.057 |
| THO1 | 0.010 | <0.056 | 0.009 | <0.057 |
| D21S11 | 0.154 | 0.114 | 0.105 | 0.058 |
| D18S51 | 0.207 | 0.341 | 0.054 | 0.175 |
| Penta E | 0.138 | 0.222 | 0.050 | 0.170 |
| D5S818 | 0.139 | 0.056 | 0.024 | <0.057 |
| D13S317 | 0.138 | 0.111 | 0.041 | <0.057 |
| D7S820 | 0.125 | 0.111 | 0.015 | 0.057 |
| D16S539 | 0.113 | 0.111 | 0.022 | <0.056 |
| CSF1PO | 0.145 | 0.111 | 0.024 | <0.057 |
| Penta D | 0.074 | 0.167 | 0.218 | <0.056 |
| vWA | 0.305 | 0.488 | 0.030 | <0.066 |
| D8S1179 | 0.162 | 0.390 | 0.019 | 0.057 |
| TPOX | 0.014 | 0.056 | 0.004 | <0.057 |
| FGA | 0.299 | 0.507 | 0.059 | 0.065 |

The mutation data obtained for the Chinese population by this laboratory is compared with those published by the American Association of Blood Banks [8] on data accumulated from European and American laboratories in Table 2.

Where mutations are observed in the Chinese population, there seemed a good correlation between the two sets of data, in particular on paternal mutation rates. The mutation rates were generally of the same order of magnitude with variations ranging from 1 to 4 fold.

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