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MUTATION OF STR IN PATERNITY TESTING

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Abstract

Since the founding of DNA fingerprint by Alec J Jeffreys in 1985, DNA analysis was widely applied in paternity testing. Nowadays, Short Tandem Repeats (STR) is the most popular DNA typing for paternity testing because of its high discrimination power, especially when the typing is performed in combination of 6, 9, 13 or 15 STR loci. STR is the nuclear DNA, and inherited from the mother and father according to Mendelian law. Every child has a pair of DNA fragment, one inherited from the mother (maternal fragment), and the other from the father (paternal fragment). In paternity testing we compare the DNA typing of the child and mother to find the maternal fragment. The other fragment of the child must be the paternal fragment. This paternal fragment of the child, then is compared to the fragments of alleged father. The result of this comparison is either match (the paternal fragment is the same as one of the alleged father's DNA fragments) or exclusion (the paternal fragment is not the same with any of the alleged father's fragments). A child IS the biological child of alleged father if in every STR locus the comparison is match. A child IS NOT the biological child of alleged father if in 2 or more STR loci the comparisons are exclusion. Single exclusion in a paternity testing, that still be a single exclusion after additional STR loci analysis is usually caused by mutation. Mutation on STR locus will causes the repeat of a person shift one step more or less than the original. In the case of mutation, the paternity index will decrease although we still confirm that the alleged father is the biological father of the child. In this paper we report 2 paternity cases that showed mutation in STR typing.

Key words: paternity testing – DNA – STR - mutation

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Introduction

Since the founding of DNA fingerprint by Alec J Jeffreys in 1985 (1), DNA analysis was widely applied in paternity testing (2,3,4,5,6,7,8). Nowadays, Short Tandem Repeats (STR) is the most popular DNA typing for paternity testing because of its high discrimination power, especially when the typing is performed in combination of 6, 9, 13 or 15 STR loci. STR is the nuclear DNA, and inherited from the mother and father according to Mendelian law. Every child has a pair of DNA fragment, one inherited from the mother (maternal fragment), and the other from the father (paternal fragment) (9).

In paternity testing we compare the DNA typing of the child and mother to find the maternal

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fragment. The other fragment of the child must be the paternal fragment. This paternal fragment of the child, then is compared to the fragments of alleged father. The result of this comparison is either match (the paternal fragment is the same as one of the alleged father's DNA fragments) or exclusion (the paternal fragment is not the same with any of the alleged father's fragments) (10). A child IS the biological child of alleged father if in every STR locus the comparison is match. A child IS NOT the biological child of alleged father if in 2 or more STR loci the comparisons are exclusion.

Sometime in real case, a mutation case is happened. Mutation in STR loci happens when there is only one unmatched (exclusion) in one locus among the loci examined, and still like that after some locus are added to examine. The other sign of mutation, is that the difference length between the alleles is only one repeat. In this condition, the probability of the paternity will be decreased. In this paper, we will report a case of mutation is paternity case

Case Report

A man, 50 years old has remarried a young lady, 25 years old. One year after their marriage, the second wife was pregnant and then delivered a baby. From the first wife, the man had already had 2 children, however since the last 10 years the husband had suffered from impotent due to uncontrolled Diebetes Melitus. The examination of semen showed relatively unfertile due to oligozoospermia. The paternity testing was performed by the request of first wife, and the result as shown in the Table 1.

Table 1. The result of DNA typing on 9 STR loci

No	DNA Locus	"Father"	Child	Mother	Conclusion
1	FGA	22, 23.2	23, 23.2	23, 23	match
2	vWA	14, 15	14, 17	17, 18	match
3	D3S1358	15, 18	15, 16	16, 17	match
4	D18S51	16, 17	14, 18	14, 15	**exclusion**
5	D21S11	30, 32	31, 32	31, 31.2	match
6	D8S1179	13, 14	12, 14	12, 15	match
7	D7S820	8, 10	8, 11	11, 12	match
8	D13S317	9, 12	11, 12	9, 11	match
9	D5S818	13, 14	12, 14	10, 12	match

This result of DNA typing is inconclusive, because there was only one exclusion from 9 DNA loci. In order to get more accurate and conclusive analysis, 6 more STR loci was examine and the result is shown in Table 2.

Table 2. The STR analysis in 6 other loci, in addition to the former analysis.

No	DNA locus	'Father"	Child	Mother	Conclusion
10	CSF1PO	11, 12	11, 11	11,11	match
11	D2S1338	19, 21	19,22	20, 22	match
12	D16S539	10, 12	10, 12	10, 12	match
13	THO1	7, 9	7, 7	7, 8	match
14	TPOX	8, 11	8, 9	8, 9	match
15	D19S433	14, 14	13, 14	13, 15.2	match

In this case, we found that (1) there were only single exclusion in a paternity testing on 9 STR loci, (2) that still be a single exclusion after additional 6 more STR loci analysis, and (3) The only un-match allele was that from D18S51 locus, 17 allele in the father and 18 allele in the child (with only 1 base different). Based on that findings, we concluded that the paternity was approved, but the paternity index was calculated low, because there was mutation in this case.

Conclusion

Mutation on STR locus will cause the repeat of a person shift one step more or less than the original. In the case of mutation, the paternity index will decrease although we still confirm that the alleged father is the biological father of the child.

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