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Non-exclusion paternity case with a triple genetic incompatibility☆

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Abstract. This work presents a paternity determination case in which the total analysis of 30 DNA markers detected three genetic inconsistencies between the putative father and the child, two in STR markers, D8S1179 and D18S51, and one in RFLP marker, D7S21. The paternity index, including the mutation rates of the systems mentioned above, amounted to 2.374×10^{10}, which corresponds with the paternity probability higher than 99.99999999%. The case proves that great care should be taken when the non-fatherhood status is ascertained on the basis of the ''three exclusion rule''. It suggests also that the best solution is to calculate the appropriate statistical estimations in every case.

Keywords: Paternity investigation; Poland; Triple mutation; D8S1179; D18S51; D7S21

1. Introduction

A mutation event of DNA markers caused by losses or gains repetitive units is quite a common phenomenon in forensic practice. It must be taken into careful consideration in genetic paternity determination. With the increase in the number of genetic loci investigated, the presence of a single genetic inconsistency between a child and his biological father has become relatively frequent and cases with double paternal genetic inconsistencies have been reported more and more often [1,2]. The aim of this paper was to show a non-exclusion paternity case with three paternal genetic incompatibilities found in our material.

2. Materials and methods

The case was encountered during routine paternity tests. Samples of blood were taken from the alleged father, his child and the child's mother. The DNA was isolated with the use of the salt extraction procedure as described by Lahiri [3]. Minisatellite analysis was

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performed by restriction with *Hin*fI and hybridization with MS43A, G3, YNH24, TBQ7, MS1, MS205, MS31 (Cellmark Diagnostics) and measurement of restriction fragments by software BIO1D (Vilber Lourmat). The amplification of STR markers was performed using the Identifiler system (Applied Biosystem) and Power Plex 16 system (Promega) with a detection on 310 ABI Prism and ABI Prism 377 sequencers. The additional six loci included in AmpliTypePM + DQA1 Typing Kit (Applied Biosystem) were co-amplified and typed utilizing reverse dot-blot.

Paternity index (PI) was calculated including the mutation rates according to Brenner [4].

3. Results

The results observed in a paternity case with three inconsistencies between the father and the child with the paternity index (PI) value are given in Table 1. The combined

Table 1
A non-exclusion paternity case with three paternal genetic inconsistencies in D8S1179, D18S51 and D7S21 loci

L.p.	Locus	Father	Child	Mother	PI
1	D8S1179	13–13	14–15	10–15	0.004[a]
2	D16S539	9–13	9–11	11–12	8.929
3	D21S11	29–30	28–30	28–30.2	2.315
4	vWA	18–19	16–18	15–16	1.984
5	TH01	7–8	6–7	6–8	4.032
6	D18S51	15–15	15–16	14–15	0.001[a]
7	D13S317	11–12	12–12	9–12	1.953
8	D7S820	9–10	10–10	10–10	1.666
9	CSF1PO	10–11	11–13	12–13	2.193
10	D19S433	12–13	13–13	13–16	2.451
11	D2S1338	20–20	20–24	16–24	6.579
12	Penta D	10–11	11–14	10–14	3.215
13	D3S1358	16–17	14–17	14–14	3.205
14	FGA	19–25	19–23	20–23	7.353
15	Penta E	9–13	13–14	12–14	2.933
16	D5S818	12–13	13–14	11–14	4.032
17	TPOX	11–11	8–11	8–8	3.968
18	LDLR	BB	BB	AB	0.806
19	GYPA	AB	AB	AA	1.000
20	HBGG	AA	AA	AB	0.960
21	D7S8	AB	AB	AB	1.000
22	GC	AA	AC	CC	2.083
23	DQA1	2–4.1	1.1–2	1.1–4.2/4.3	5.618
24	D12S11	10.4–3.5 kb	10.4–7.4 kb	10.2–7.4 kb	50.000
25	D7S22	10.5–4.6 kb	10.5–2.9 kb	9.3–2.9 kb	25.000
26	D2S44	4.5–1.9 kb	4.5–2.7 kb	3.3–2.7 kb	12.500
27	D10S28	4.7–2.0 kb	4.7–3.0 kb	3.0–1.4 kb	14.286
28	D1S7	9.7–2.3 kb	9.7–6.8 kb	8.3–6.8 kb	15.625
29	D16S309	3.4 kb	3.4–2.6 kb	2.9–2.6 kb	10.526
30	D7S21	8.4 kb	8.9–7.6 kb	7.6–5.9 kb	0.264[a]
Total					2.374×10^{10}

PI—paternity index value.
[a] Value of PI with consideration of appropriate average paternal mutation rates.

paternity index in the case after inclusion of the mutation index for D8S1179, D8S51 and D7S21 was 2.374×10^{10}, which corresponded with final probability of paternity value higher than 99.99999999%. From this statistical calculation, we concluded the presence of a triple mutation event in the investigated case.

4. Discussion

Though the triple mutation theoretically seems to be an extremely rare incident, it does not appear impossible in our practice. Minisatellites such as D7S21 are considerably mutable regions in the human DNA with an average paternal mutation rate of 1.9×10^{-2} and the lack of maternal mutation event [5]. For STR markers, an average paternity mutation rate of 3.4×10^{-3} is 10 times higher than the maternal one [1]. Taking into consideration mentioned values of mutation rates, combined frequency of observed triple mutation event appears in 1 out of 4.5 million cases on average, as previously reported Thomson and Pilotti [6].

5. Conclusion

We proved that ‘‘three exclusion rule’’ is not safe enough. We suggest to rule out any arbitrary decision point and to calculate the appropriate statistical estimations in every case.

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