



## The most frequent autosomal STRs involved in exclusion of paternity cases in a population from southeast, Mexico

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### ABSTRACT

**Introduction:** Short tandem repeats (STR) are very widespread in the human genome; being a source of polymorphic markers, used for the determination of kinship. Few studies indicate the frequency of autosomal STR involved in the exclusion of paternity, as well as, those with a higher percentage of discordance. determine the distribution and frequency of the main autosomal STR markers involved in the exclusion of paternity cases in an admixed population from Southeast, Mexico (Peninsula de Yucatán).

**Methods:** 91 cases of excluded paternity, were included in the study: 61 trios and 30 motherless. All subjects belong to an admixed population with Mayan ethnicity from Southeast of Mexico. PowerPlex Fusion System (PPFS) was used for genotyping. The distribution and frequency of the autosomal STRs involved in each case of paternity exclusion were recorded based on 22 autosomal STR of PPFS.

**Results:** The number of STR which determined the exclusion in all cases ranged from 5 to 18 markers, being up to 14 STR for motherless cases. Paternity exclusions occurred most frequently with 13-STRs (22.22%) for trios and 9-STRs (37.5%) for motherless cases. The autosomal STR with the highest percentage of discordance is PENTA-E, for all cases 69.23%, 85.25% for trios. For motherless cases were D1S1656 and PENTA-E (36.67%, both).

**Conclusions:** The most frequent number of autosomal STR marker to confer a paternity exclusion was 13, the most informative STR marker for exclusions was PENTA-E.

### 1. Introduction

The human genome contains thousands upon thousands of STR markers, only a small core set of loci have been selected for use in forensic DNA and human identity testing [1]. According to the recommendation of FBI the CODIS Core Loci are twenty: CSF1PO, D3S1358, D5S818, D7S820, D8S1179, D13S317, D16S539, D18S51, D21S11, FGA, TH01, TPOX, vWA, D1S1656, D2S441, D2S1338, D10S1248, D12S391, D19S433, D22S1045 [2]. These loci are commonly used to paternity testing largely because of their ease of use in the form of commercial STR kits [3]. Commercial kits as PowerPlex Fusion include the main 20 autosomal STR markers and two pentanucleotide: PENTA D and PENTA E.

Few studies indicate the frequency of autosomal STR involved in the exclusion of paternity, as well as, those with a higher percentage of discordance. New genetic systems with a higher number of STRs are poorly studied in Mexican Mestizo populations, so, in this study, we describe the distribution and frequency of the main autosomal STR

involved in the exclusion of paternity cases in an admixed population from Southeast of Mexico (Peninsula de Yucatán).

To determine the distribution and frequency of the main autosomal STR markers involved in the exclusion of paternity cases in an admixed population from Southeast of Mexico (Peninsula de Yucatán).

### 2. Methods

Information dataset was obtained from 334 paternity cases (exclusion and inclusion) performed over five years (2015–2019) by a private Mexican laboratory ([www.dimygen.com](http://www.dimygen.com)). We selected (N = 91) cases of excluded paternity for the study: 61 trios and 30 motherless. All subjects belong to an admixed population with Mayan ethnicity from Southeast of Mexico. Inform consent was signed by each participant.

The DNA extraction was carried out with the Swab Solution System (Promega Corp.) from buccal swabs. PowerPlex® Fusion Kit (PPF) (Promega) was used for genotyping, PCR fragments were amplifying from Thermal Cycler ARKTIK® (Thermo Scientific). Amplified products

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**Table 1**  
Number of mismatches obtained from exclusion of paternity cases, trios and motherless.

Total STRs exclusion	Trio		Motherless		All cases	
	N	%	N	%	N	%
5	0	0.00	1	3.33	1	1.10
6	0	0.00	1	3.33	0	0.00
7	1	1.64	1	3.33	2	2.20
8	1	1.64	3	10.00	4	4.40
9	1	1.64	11	36.67	12	13.19
10	5	8.20	4	13.33	9	9.89
11	5	8.20	3	10.00	8	8.79
12	8	13.11	3	10.00	11	12.09
13	14	22.95	2	6.67	16	17.58
14	8	13.11	1	3.33	9	9.89
15	7	11.48	0	0.00	7	7.69
16	4	6.56	0	0.00	4	4.40
17	6	9.84	0	0.00	6	6.59
18	1	1.64	0	0.00	1	1.10
Total	61	100	30	100	91	100

**Table 2**  
Frequency of exclusions per each STR paternity for trio and motherless.

Loci	All cases		Trio		Motherless	
	N	%	N	%	N	%
PENTA E	63	69.23	52	85.25	11	36.67
D18S51	60	65.93	50	81.97	10	33.33
D1S1656	56	61.54	45	73.77	11	36.67
FGA	53	58.24	47	77.05	6	20
D13S317	51	56.04	45	73.77	6	20
D2S1338	50	54.95	40	65.57	10	33.33
D21S11	49	53.85	41	67.21	8	26.67
D12S391	49	53.85	43	70.49	6	20
D19S433	47	51.65	39	63.93	8	26.67
PENTA D	44	48.35	37	60.66	7	23.33
vWA	41	45.05	36	59.02	5	16.67
D5S818	41	45.05	36	59.02	5	16.67
D8S1179	40	43.96	32	52.46	8	26.67
TPOX	38	41.76	34	55.74	4	13.33
D16S539	36	39.56	30	49.18	6	20
D3S1358	35	38.46	31	50.82	4	13.33
D10S1248	35	38.46	31	50.82	4	13.33
D7S820	35	38.46	32	52.46	3	10
D2S441	33	36.26	29	47.54	4	13.33
TH01	33	36.26	28	45.9	5	16.67
CSF1PO	30	32.97	27	44.26	3	10
D22S1045	27	29.67	23	37.7	4	13.33

were analyzed by capillary electrophoresis with the ABI Prism™ 310 (Applied Biosystem). The Genemapper software V.5 was used to allocation alleles. The distribution and frequency of the autosomal STRs involved in each case of paternity exclusion were recorded based on 22 autosomal STR of PPF. According to the recommendations on biostatistics in paternity testing by Gjertson et al [4], the criteria for exclusion of paternity was determined when more than two mismatches were detected.

Descriptive statistics was applied to the dataset recollected captured in a Microsoft Excel spreadsheet. Type of cases were classified by trio and motherless. Then, reported the number of loci detecting the exclusion and the frequent of autosomal STR involved in the exclusion of paternity.

**3. Results**

From 334 paternity cases performed, we observed 91 cases of paternity exclusions, resulting in 27.25% testing exclusion rate. Trios were the most common test (61 cases, 67.7%), followed by motherless

(30 cases, 32.3%) (Table 1).

The Number of STR loci which determined the exclusion (mismatches) in all cases ranged from 5 to 18, being up to 14 STR for motherless cases. Paternity exclusions occurred most frequently with 13-STRs (22.95%) for trios and 9-STRs (36.67%) for motherless cases (Table 1).

The most frequent autosomal STR of discordance was PENTA-E, for all cases 69.23% and for trio 85.25%. For motherless cases were D1S1656 and PENTA-E (36.67%, both). (Table 2).

**4. Discussion**

We obtained 27.5% of paternity testing exclusion rate in this study, which is similar to the exclusion rate (29.58%) for Mexican population in 3005 paternity test cases [5] and consistent to the range of exclusion rate reported by the American Association of Blood Banks (AABB) from 4.5 to 33% [6].

In our dataset trio was the most common paternity test modality in this population. Opposite situation was reported by a previous study in Mexico where motherless cases are the most common tests (77.27%), followed by trios including the mother (20.7%). In the study, they reported this options were preferred probably by the following reasons: i) it is cheaper that when mother is included (trio); ii) the father does not want that the mother realize about it; iii) mother's participation is not obligatory because the large majority of the tests are for personal (no legal) purposes [5]. One of the strongly reasons in our study probably who explain why trio cases where the most frequently is due to the cost is the same for trio or motherless; in order to increase the scientific power of the test. Therefore, if the mother is available or agree to participate it will not be an additional cost. For all cases from dataset did not discriminate by legal or personal modality.

The average of the number of STR which determined the exclusion of paternity test in motherless cases was 9.58, which is within the range reported by Garcia-Aceves et al. 2018 [5] in the Mexican population from 8.83 to 10.65 for father-daughter and father-son cases, respectively. Our results do not discriminate father-daughter or father-son cases.

The most frequent number of autosomal STR marker to confer a paternity exclusion was 13 for all cases and trios and 9 for motherless. The most informative STR marker for exclusions was PENTA-E for all cases, both trios and motherless being within the range reported in the same Mexican previous study. In general, the estimated STR exclusion rate, as well as the average number of loci to paternity exclusion test; are in agreement with previous reports.

**5. Conclusion**

This is the first study who reports results of paternity exclusions test in a population from Southeast of Mexico. It stands out that trio was the most frequent (61%) paternity testing. The paternity exclusion rate was 27.5%. The most frequent number of autosomal STR marker to confer a paternity exclusion was 13. The most informative STR marker for exclusions was PENTA-E.

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