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The implementation of Alu insertion polymorphism as a genetic marker for forensic investigation in a Jordanian population sample

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Abstract

Alu element is one of the most abundant short interspersed nuclear elements (SINE) in the human genome with great forensic potential. The current study focused on the analysis of the utility of *Alu* insertion polymorphism in forensic DNA typing in a Jordanian population sample (central Jordan). *Alu* insertions in seven different genetic loci were amplified using Polymerase Chain Reaction (PCR). The frequencies of the *Alu* insertions were 0.345 for *ACE*, 0.441 for *TPA25*, 0.291 for *PV92*, 0.845 for *APO*, 0.468 for *FXIIIB*, 0.727 for *HS3.23* and 0.527 for *B65*. *APO* and *FXIIIB Alu* insertions were the most dominant alleles, while *PV92 Alu* insertion was the least frequent. Combined Power of Discrimination (P_D) for those seven loci was 0.998285646, whereas the combined Power of Exclusion (P_E) was 0.4987.Combining the seven *Alu* insertion/deletion data along with the Jordanian combined STR data should give an exceptional resolution power between Jordanian individuals.

Keywords: *Forensic science, Alu polymorphism, Allelic frequency, Jordanians*

1. Introduction

The implementation of DNA fingerprinting of different DNA sequences such as variable number tandem repeats (VNTR), short tandem repeats (STRs) and single-nucleotide polymorphisms (SNPs) in criminal situations has tremendously advanced the field of forensic investigation. STRs increased the capability of solving questionable situations through their extremely high power of identification or discrimination and paternity testing due to their polymorphic nature (Butler, 2002; and Alonso *et al.*, 2018). Though STRs capabilities are still undisputed, many investigators in recent years have studied other polymorphic DNA sequences. For example, the dimorphic *Arthrobacter luteus* restriction endonuclease (*Alu*) mobile elements are studied for potential use in population genetic studies and forensic investigations in order to increase the certainty of juridical perceptiveness (Gill, 2001; Frudakis *et al.*, 2003; Petkovski *et al.*, 2003; and Terreros *et al.*, 2009).

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The *Alu* elements are approximately 300 bp in length and constitute one of the most plentiful short interspersed nuclear elements (SINE) with more than one million copies accounting for more than 10% of the mass of the human genome (Lander *et al.*, 2001; Deininger and Roy-Engel, 2002; Batzar *et al.*, 2002; and Ray *et al.*, 2007). They are mostly fixed which means that at a particular locus all individuals are homozygous for *Alu* insertion (Ray *et al.*, 2007). Furthermore, it has been estimated that they can insert themselves in the human genome at a rate of 100 to 200 copies per million years via a process known as retro insertion (Batzar *et al.*, 1994). Yet, no mechanism for complete *Alu* element removal after insertion is known (Roy-Engel *et al.*, 2002).

Some of these insertions are still polymorphic for presence or absence at a specific location due to recent transposition in the human genome, and the absence of the insertion is considered as the ancestral state and presumably, the gain of the *Alu* element is the mutational change (Stoneking *et al.*, 1997). The ability of the *Alu* elements to determine the ancestral state and the direction of mutational changes renders them as unique markers in population genetic studies. In this context, *Alu* polymorphism is widely considered as a tool in evolutionary, population's ethnicity and origin, and genetic diversity studies among populations (Batzar *et al.*, 1996; Roy-Engel *et al.*, 2001; Tripathi *et al.*, 2017; Bahri *et al.*, 2011; and Zanetti *et al.*, 2014). It has been proved that it is a useful tool for inference of human geographic origins (Ray *et al.*, 2007). In addition, some of these elements are present in large frequencies in some populations and are identified as "population specific alleles" so they can be used as a differential marker between two well-defined populations (Shriver *et al.*, 1997).

Many investigators have reported the potential use of *Alu* elements in forensic identification such as paternity testing. Chadli and coworkers (2009) demonstrated the forensic importance of seven loci which have the insertion or deletion of *Alu* fragment in south Morocco. Studies on the German, Japanese, Taiwanese and other populations showed that *Alu* polymorphisms provide a useful tool in forensic DNA analysis where *Alu* repeats might be unique to an individual and thus they can provide information on human population relationships (Wallace *et al.*, 1991; Batzar *et al.*, 1996; Schneider *et al.*, 1996; Hsieh *et al.*, 2002; and Asari *et al.*, 2012).

In the current study, we focused on amplifying *Alu* insertion on seven different loci including Angiotensin Converting Enzyme (*ACE)*, Tissue Plasminogen Activator (*TPA25)*, Apolipoprotein (*APO)*, Improved Coagulation Factor XIIIB (*FIIIXB)*, *Alu* Insertion PV92 (*PV92)*, Human Specific 3.23 (*HS3.23)*, and HLA-B65 (*B65)*. The variation in allele and genotype frequencies was studied in a Jordanian population sample residing in central Jordan to test the potential use of these loci as genetic markers for identity testing.

2. Materials and Methods

2.1. Sample collection and DNA isolation

Blood samples were collected in EDTA tubes from 110 unrelated Jordanian individuals residing in central Jordan, including 55 men and 55 women. Informed consent was obtained from each individual, and the study was approved by the Institute Review Board (IRB) of Hashemite University, which conforms to the World Medical Association Declaration of Helsinki. DNA was isolated using the Wizard® Genomic DNA Purification Kit (Promega, Madison, USA). DNA samples were quantified using spectrophometry (Biowave II, Biochrom, UK) and stored at –80 °C until further use.

2.2. DNA amplification using PCR

Alu fragment on seven loci including *ACE*, *TPA25*, *PV92*, *APO*, *FXIIIB*, *HS3.23*, and *B65* were amplified using the Polymerase Chain Reaction (PCR). Fourteen specific primers for the flanking *Alu* insertion sequence (Table 1) were used for site amplifications (Chadli *et al.*, 2009). The PCR amplification was carried out using

Table 1: Locus characteristics, primer sequences, annealing temperatures, and PCR product size for genetic *Alu* elements in seven loci

Locus / Position	Forward primer	Reverse primer	Annealing temperature	Product size (bp)
ACE17q23	CTGGAGACCACTCCCA TCCTTTCT	GATGTGGCCA TCACATTCGTCAGAT	58 °C	480-191
TPA258p11.2	GTAAGAGTTCCGTAAC AGGACAGCT	CCCCACCCTAGGAG AACTTCTCTTT	58 °C	400-110

Table 1 (Cont.)

Locus / Position	Forward primer	Reverse primer	Annealing temperature	Product size (bp)
PV92 16q24.2	AACTGGGAAAATTTG AAGAGAAAGT	TGAGTTCTCAACTC CTGTGTGTTAG	54 °C	437-122
APO 11q23-q24	AAGTGCTGTAGGCCAT TTAGATTAG	AGTCTTCGATGACA GCGTATACAGA	50 °C	409-96
FXIIIB 1q31-q32.1	TCAACTCCATGAGATT TTCAGAAGT	CTGGAAAAAATGTA TTCAGGTGAGT	56 °C	720-450
HS3.237	GGTGAAGTTTCCAACG CTGT	CCCTCCTCTCCCTTT AGCAG	52 °C	410-110
B6511q14.2	ATATCCTAAAAGGGA CACCA	AAAATTTATGGCAT GCGTAT	60 °C	420-81

100 ng DNA, 1.5 mM $MgCl_2$, 0.2 mM dNTP, 1 unit of *Taq* polymerase (Bio Basic, Canada) and 0.24 pM of each primer (GeneWiz, USA). The PCR protocol involved 35 cycles of repeated denaturation of DNA template at 94 °C for 1 min, annealing of primers for 2 min as described previously (Chadli *et al.*, 2009), and DNA synthesis at 72 °C for 2 min. The electrophoresed PCR products were visualized on 2% agarose gel stained with RedSafe™ Nucleic Acid Staining Solution (iNtRON Biotechnology DR, USA) under UV.

3. Statistical Analysis

Allelic and genotypic frequencies were calculated according to the counting method. Observed (H_O) and Expected (H_E) Heterozygosity, Power of Identity (P_I), Power of Discrimination (P_D), Power of Exclusion (P_E), Polymorphic Information Content (PIC), Combined or Joined P_I, P_D and P_E were calculated according to the formulas described previously (Brenner and Morris, 1990; Evett and Weir, 1998; and Chesnokov and Artemyeva, 2015). The observed genotypes and alleles frequencies were compared with those expected in order to verify the Hardy-Weinberg Equilibrium (HWE). The chi-square test and Fisher's exact test were performed for the polymorphism frequency. Statistical analysis for allele interpopulation analysis was performed using Statistica software, StatSoft Inc, Tulsa, OK, USA (version 10). A value of $p < 0.05$ was considered statistically significant.

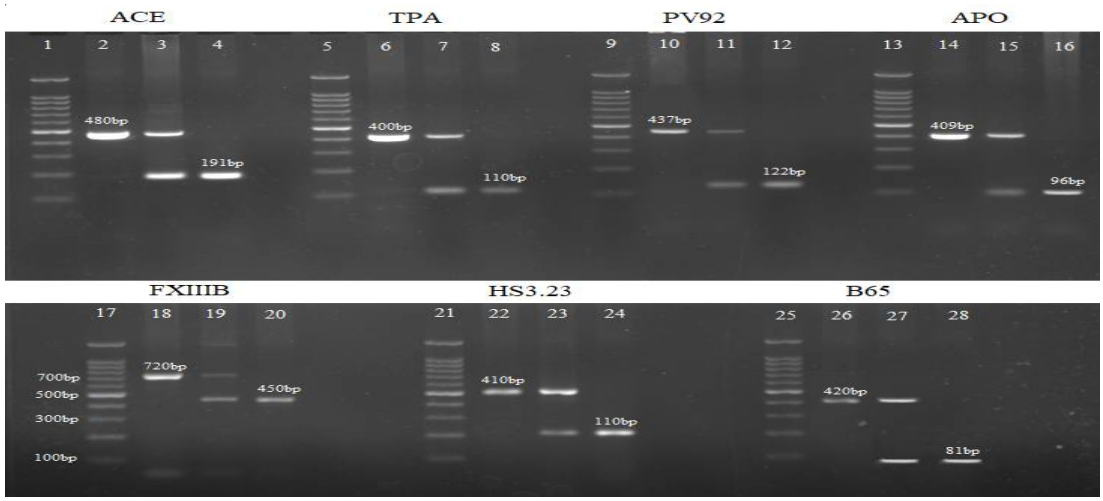

Figure 1: Representative PCR amplification products of the seven *Alu* DNA genetic loci from unrelated Jordanian blood samples

Note: Lanes 1, 5, 9, 13, 17, 21, and 25: 100 bp ladder. The rest of the lanes represents Alu DNA polymorphism for the seven loci with their respective molecular size in base pair.

4. Results and Discussion

Alu DNA fragment exhibits a dimorphic nature in which its polymorphism depends on its stable integration or absence at a certain locus (Sawada *et al.*, 1985; Sawada and Schmid, 1986; Bailey and Shen, 1993), and the rare loss of such insertions (Edwards and Gibbs, 1992). It has been speculated that the *Alu* elements have a copy number of over a million per genome (Edwards and Gibbs, 1992) with potential impacts in human evolution and forensics. In the current study, *Alu* insertions in seven loci (including *ACE, TPA25, PV92, APO, FXIIIB, HS3.23* and *B65*) were analyzed for their prospective forensic implications in a Jordanian population sample (Figure 1).

The genotypic and allelic frequencies of these loci and the evaluation of the HWE in the sample population are shown in Table 2. The frequency of the *Alu* insertion were 0.345 for *ACE*, 0.441 for *TPA25*, 0.291 for *PV92*, 0.845 for *APO*, 0.468 for *FXIIIB*, 0.727 for *HS3.23* and 0.527 for *B65*. For *APO* and *FXIIIB*, *Alu* insertion (Alu^+) was the predominant allele with a frequency of 0.845 and 0.727 respectively, while the *PV92 Alu* insertion was the least with a frequency of 0.291. Chi-square test statistics values showed no significant departure from HWE for all of the seven loci. Results have also shown no significant difference in *Alu* insertion allelic distribution between Jordanian males and females ($p > 0.05$) (data not shown). Furthermore, no significant difference between observed and expected heterozygosity was also observed ($p > 0.05$). *ACE* and *FXIIIB* locus have showed the higher heterozygosity values (0.4181 and 0.4272 respectively) which indicates that the Jordanian population is variable in regards to these loci. On other hand, *APO* loci have showed the lower heterozygosity (0.2181).

The P_D was calculated by locus and for all of the studied loci (Table 2). P_D was 0.608 for *ACE*, 0.6598 for *TPA25*, 0.576 for *PV92*, 0.408 for *APO*, 0.652 for *FXIIIB*, 0.562 for *HS3.23* and 0.664 for *B65*. *APO* and *FXIIIB Alu* insertions were respectively the most dominant alleles, while *PV92 Alu* insertion was the least frequent. *B65 Alu*

Table 2: Genotypic, allelic frequencies and forensic indices of the insertions and deletions of the *Alu* DNA fragment per seven genomic loci in 110 unrelated Jordanian individuals

Forensic parameters		*Alu* insertion/deletion frequency *per* locus						
		ACE	***TPA25***	***PV92***	***APO***	***FXIIIB***	***HS3.23***	***B65***
Genotype	$Alu^{+/+}$	0.1363	0.2818	0.1000	0.7363	0.2554	0.5454	0.3454
	$Alu^{-/-}$	0.4181	0.3181	0.3818	0.2181	0.4272	0.3636	0.3636
	$Alu^{+/-}$	0.4454	0.4000	0.5181	0.0454	0.3181	0.0909	0.2909
Allele	Alu^{+}	0.3450	0.4410	0.2910	0.8450	0.4680	0.7270	0.5270
	Alu^{-}	0.6550	0.5590	0.7090	0.1550	0.5320	0.2730	0.4730
H_O		0.4181	0.3181	0.3818	0.2181	0.4272	0.3636	0.3636
H_E		0.4520	0.4930	0.4126	0.2620	0.4980	0.3969	0.4985
p-value		0.9601	0.8034	0.9617	0.9314	0.9199	0.9578	0.8475
PIC		0.4520	0.4930	0.4130	0.2620	0.4980	0.3970	0.4990
P_I		0.3920	0.3402	0.424	0.5920	0.3480	0.4380	0.336
P_D		0.6080	0.6598	0.5760	0.4080	0.6520	0.5620	0.6640
P_E		0.1260	0.0710	0.1030	0.0350	0.1310	0.0940	0.0940
CP_I		0.001714354						
CP_D		0.998285646						
CP_E		0.4987						

Note: ($^{+/+}$): Homozygous insertion; ($^{+/-}$): Heterozygous insertion / deletion; ($^{-/-}$): Homozygous deletion; ($^{+}$): *Alu* insertion; ($^{-}$): *Alu* deletion; H_O: Observed heterozygosity; H_E: Expected heterozygosity; PIC: Polymorphic Information Content; P_I: Power of Identity; P_D: Power of Discrimination; P_E: Power of Exclusion; CP_I: Combined Power of Identity; CP_D: Combined Power of Discrimination; and CP_E: Combined Power of Exclusion.

insertion displayed the highest P_D (0.664) with a P_I value of 0.336, while the *APO Alu* insertion exhibited the lowest P_D of 0.408. Therefore, *APO* is the least powerful locus and *B65* is the most powerful loci among the seven loci studied. The combined P_D for the seven loci included in this study was 0.998285646. P_E ranged from 0.035 for *APO* and 0.131 for *FXIIIB* (Table 2), and the combined P_E for the seven loci included in this study was 0.4987.

In addition, the discrepancy in *Alu* insertion frequency between Jordanians and other populations was analyzed (Table 3). The data showed a significant difference in *Alu* insertion allele frequency between Jordanians

Table 3: Comparison of *Alu* insertion frequencies between Jordanians and 38 human populations

Population – *p*-value	***TPA25***	***PV92***	***APO***	***ACE***	***FXIIIB***	***B65***	***HS3.23***
Jordan (Current study)	0.441	0.291	0.845	0.345	0.468	0.527	0.727
African American	0.302	0.209	0.57	0.488	0.221	-	0.99
p-value	**0.0419**	0.1806	**<0.0001**	**0.0403**	**0.0002**	**-**	**<0.0001**
Afro-Caribbean	0.286	0.143	0.5	0.524	0.31	-	-
p-value	**0.0227**	**0.0111**	**<0.0001**	**0.0107**	**0.0219**	-	-
Alaska Natives	0.298	0.619	0.917	0.583	0.917	-	-
p-value	**0.0362**	**<0.0001**	0.1159	**0.0007**	**<0.0001**	-	-
Armenians	0.43	0.013	0.871	0.477	0.343	0.453	-
p-value	0.8753	**<0.0001**	0.5984	0.0578	0.0718	0.2592	-
Azerbaijanis	0.513	0.382	0.943	0.216	0.1	0.697	-
p-value	0.3081	0.1733	**0.0244**	**0.0423**	**<0.0001**	**0.0136**	-
Benin	0.463	0.225	0.488	-	-	0.763	-
p-value	0.7546	0.2861	**<0.0001**	-	-	**0.0005**	-
Bretons	0.556	0.267	0.9	0.478	0.4	-	-
p-value	0.1039	0.7051	0.2436	0.0560	0.3320	-	-
Cambodian	0.5417	1	0.7917	-	-	0.4167	-
p-value	0.1544	**<0.0001**	0.3283	-	-	0.1182	-
Cameron	0.343	0.343	0.75	-	-	0.625	-
p-value	0.1558	0.4294	0.0946	-	-	0.1608	-
Cherkessians	0.386	0.167	0.932	0.39	0.439	0.651	-
p-value	0.4297	**0.0369**	0.0506	0.5093	0.6804	0.0747	-
Chinese	0.4412	0.8529	0.8824	-	-	0.4706	-
p-value	0.9977	**<0.0001**	0.4408	-	-	0.4251	-
Darginians	0.361	0.167	0.864	0.167	0.143	0.321	-
p-value	0.2484	**0.0369**	0.7032	**0.0039**	**<0.0001**	**0.0032**	-
Egyptian	0.513	0.125	0.8	-	-	0.55	-
p-value	0.3081	**0.0038**	0.4050	-	-	0.7442	-
European American	0.55	0.178	0.944	0.511	0.467	-	-

Table 3 (Cont.)

Population – *p*-value	***TPA25***	***PV92***	***APO***	***ACE***	***FXIIIB***	***B65***	***HS3.23***
p-value	0.1232	0.0593	**0.0227**	**0.0177**	0.9887	-	-
Finnish	0.4444	0.1563	0.9737	-	-	0.4211	-
p-value	0.9614	**0.0223**	**0.0015**	-	-	0.1337	-
French	0.557	0.227	0.989	0.477	0.42	0.625	-
p-value	0.1009	0.3016	**0.0002**	0.0578	0.4945	0.1608	-
French Acadians	0.433	0.178	0.922	0.511	0.478	-	-
p-value	0.9092	0.0593	0.0897	**0.0177**	0.8874	-	-
Georgians	0.493	0.25	0.934	0.354	0.61	0.727	-
p-value	0.4611	0.5140	**0.0447**	0.8938	0.0440	**0.0035**	-
Greek Cypriots	0.53	0.25	0.95	0.39	0.616	-	-
p-value	0.2080	0.5140	0.0144	0.5093	**0.0357**	-	-
Greenland Natives	0.333	0.607	0.940	0.548	0.786	-	-
p-value	0.1169	**<0.0001**	**0.0301**	**0.0039**	**<0.0001**	-	-
Hispanic	0.625	0.523	0.92	0.545	0.705	-	0.74
p-value	**0.0091**	**0.0008**	**0.0996**	**0.0044**	**0.0007**	-	0.8353
Indian	0.5957	0.4662	0.8148	-	-	0.4596	-
p-value	**0.0286**	**0.0106**	0.5698	-	-	0.3405	-
Ingushians	0.224	0.129	0.941	0.34	-	0.21	-
p-value	**0.0011**	**0.0049**	**0.0281**	0.9406	-	**<0.0001**	-
Japanese	0.5	0.8571	0.8438	-	-	0.4118	-
p-value	0.4032	**<0.0001**	0.9813	-	-	0.1024	-
Kenyan	0.363	0.45	0.613	-	-	0.625	-
p-value	0.2606	**0.0199**	**0.0002**	-	-	0.1608	-
Malays	0.25	0.5	0.8333	-	-	0.5	-
p-value	**0.0045**	**0.0025**	0.8218	-	-	0.7025	-
Moroccan	0.4550	0.1961	0.6974	0.1760	0.2670	0.3727	0.8038
p-value	0.8422	0.1180	**0.0130**	**0.0065**	**0.0032**	**0.0283**	0.2000
Nigerians	0.409	0.091	0.5	0.273	0.083	-	-
p-value	0.6471	**0.0003**	**<0.0001**	0.2706	**<0.0001**	-	-
Northern Europe	0.5522	0.2537	0.9697	-	-	0.6186	-
p-value	0.1158	0.5535	**0.0024**	-	-	0.1904	-
Polish	0.75	0.1250	0.95	-	-	0.35	-

Table 3 (Cont.)

Population – *p*-value	*TPA25*	*PV92*	*APO*	*ACE*	*FXIIIB*	*B65*	*HS3.23*
p-value	**<0.0001**	**0.0038**	**0.0144**	-	-	**0.0117**	-
Pygmies	0.221	0.309	0.794	0.221	0.015	-	-
p-value	**0.0009**	0.7812	0.3484	0.0516	**<0.0001**	-	-
Rwandan	0.275	0.413	0.913	-	-	0.675	-
p-value	**0.0143**	0.0709	0.1404	-	-	**0.0326**	-
Sudanese	0.363	0.4	0.7	-	-	0.75	-
p-value	0.2606	0.1051	**0.0145**	-	-	**0.0010**	-
Swiss	0.453	0.198	0.942	0.372	0.477	-	-
p-value	0.8645	0.1260	**0.0262**	0.6905	0.8986	-	-
Turkish Cypriots	0.576	0.333	0.985	0.333	0.394	-	-
p-value	0.0562	0.5215	**0.0004**	0.8577	0.2907	-	-
Emirati	0.44	0.3	0.97	0.33	0.39	0.41	-
p-value	0.9886	0.8891	**0.0023**	0.8225	0.2651	0.0973	-
US Caucasian	-	-	-	-	-	-	0.87
p-value	-	-	-	-	-	-	**0.0117**
Vietnamese	0.2778	0.875	0.9444	-	-	0.4444	-
p-value	**0.0162**	**<0.0001**	**0.022**	-	-	0.2426	-

and Americans from different ethnicities, western Europeans, and Africans for *ACE*, *FXIIB*, *HS3.23* and *B65* ($p \leq 0.05$) (Table 3) (Arcot *et al.*, 1996; Batzer *et al.*, 1996; Nasidze *et al.*, 2001; Watkins *et al.*, 2001; and Chadli *et al.*, 2009). Furthermore, *TPA25* locus showed a significant difference in *Alu* insertion between Jordanians and Americans and Asians populations including Indians, Vietnamese, and Malay ($p \leq 0.05$), and Americans, Asians, and Europeans for *APO* and *PV92* ($p \leq 0.05$) (Arcot *et al.*, 1996; Batzer *et al.*, 1996; Nasidze *et al.*, 2001; Watkins *et al.*, 2001; Terreros *et al.*, 2005; and Chadli *et al.*, 2009). In general, the comparison between *Alu* insertion frequencies in Jordanian population and other populations [African, North and South Americans and Eastern European (African American, African Caribbean, Alaskan, Hispanic, Nigerian, Moroccan, Polish and Greenland] show a significant difference (Table 3) (Arcot *et al.*, 1996; Batzer *et al.*, 1996; Nasidze *et al.*, 2001; Watkins *et al.*, 2001; and Chadli *et al.*, 2009) ($p \leq 0.05$). These genetic variances could be due to the large geographic distance between Jordanians and those populations (Terreros *et al.*, 2009).

On the other hand, many other populations did not show a significant difference in the *Alu* insertion frequency (Armenians, Bretons, Cambodians, Cameron, Cherkessians, Chinese, Egyptians, French Acadians, Japanese, Sudanese, Swiss, Turkish Cypriots, and Emirati) when compared to Jordanians (Table 3) (Batzer *et al.*, 1996; Arcot *et al.*, 1996; Watkins *et al.*, 2001; Nasidze *et al.*, 2001; Chadli *et al.*, 2009; and Terreros *et al.*, 2005) ($p \geq 0.05$). This suggests that the similarity may be due to the occurrence of the *Alu* insertion event former to migration of modern human out of Africa 100,000 years ago, or in some cases, due to close geographic distance or the possible migratory action between these populations (Stringer and Andrews, 1988; Harpending *et al.*, 1993; Cavalli-Sforza *et al.*, 1994; Goldstein *et al.*, 1995; Nei, 1995; Stoneking *et al.*, 1997; and Terreros *et al.*, 2009).

5. Conclusion

In the light of establishing an automated fluorescent based method for *Alu* genotyping (Asari *et al.*, 2012), the researchers think that despite the small sample size included in the current study, the seven *Alu* insertion /

deletion data loci are applicable in forensic investigations and when combined with the Jordanian STR data (Yasin *et al.*, 2005) an exceptional resolution power between Jordanian individuals is maintained.

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