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Use of the Systems D1S80, D17S30, ApoB, TC11, VWA, and SE33 for Zygosity Determination - Letter to the Editor

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Abstract. The usefulness of PCR systems (D1S80, D17S30, ApoB, TC11, VWA, and SE33) is discussed. The statistical evaluation shows that these systems – together with other systems – are well suited for zygosity testing.

Key words: Zygosity testing in twins, Systems D1S80, D17S30, ApoB, TC11, VWA and SE33.

In Vol. 44 (1995: 25-30) of this journal, an article [2] discussed the use of single– and multi-locus and PCR-systems for zygosity determination. Unfortunately, the authors do not say anything about the genotypes and frequencies of the PCR-systems (D1S80, D17S30, ApoB, TC11, VWA, and SE33) whose effectiveness in zygosity determination their study tested. Moreover, the authors' conclusion that their results prove monozygosity from the fact that the twins they studied were identical in all systems, is incorrect. And their statistical interpretation is somewhat curious in its neglect of the parents' genotypes and the differing respective frequencies of the genotypes.

There are three basic methods for determining zygosity in twins; tests involving parents (cf. [1]), tests excluding parents (cf. [6]) and tests using questionnaires (cf. [7], [11]). Due to the missing parental data in the paper cited, we tested zygosity by the second method see ([6] and [9] for details of this technique) in three ways: the twins show the rarest, the most frequent or medium-frequent genotypes respectively in all systems. As can be seen from Tables 1-3, the minimum probability for monozygosity of 99.9% required by the forementioned coauthors [2] was only obtained by using the rarest genotypes, while the value obtained by using the most frequent genotypes in each system (97.27%) is below this required minumum. It is evident that these cases are very rare. Normally there will be a mixture of more or less frequent genotypes. The genotypes which occur with medium frequency result in a probability of just 98.99%. Generalizing,

Table 1 - Zygosity determination using the rarest genotypes of the systems D1S80, D17S30, ApoB, TC11, VWA and SE33*

System	Genotype	Expected frequency (%)	Monozygosity probability for each system	Bibliography of data used
D1S80	16-34	0.003	61.69	[3]
D17S30	13-13	0.002	76.13	[8]
ApoB	2-2	0.002	76.16	[8]
TC11	11-11	0.010	75.95	[4]
VWA	13-13	0.002	76.13	[5]
SE33	V12-V12	0.0004	76.23	[10]
		Combined:	99.94	

*The nomenclature of systems and genotypes is that of the authors cited.

Table 2 - Zygosity determination using the most frequent genotypes of the systems D1S80, D17S30, ApoB, TC11, VWA and SE33

System	Genotype	Expected frequency (%)	Monozygosity probability for each system	Bibliography of data used
D1S80	18-24	17.354	59.70	[3]
D17S30	2-4	12.484	60.25	[8]
ApoB	4-5	20.775	59.33	[8]
TC11	6-10	11.633	60.34	[4]
VWA	16-17	12.946	60.19	[5]
SE33	V18-V19	1.456	61.52	[10]
		Combined:	97.27	

Table 3 - Zygosity determination using medium-frequent genotypes of the systems D1S80, D17S30, ApoB, TC11, VWA and SE33

System	Genotype	Expected frequency (%)	Monozygosity probability for each system	Bibliography of data used
D1S80	18-18	6.401	67.22	[3]
D17S30	2-2	5.808	67.65	[8]
ApoB	4-4	7.673	66.38	[8]
TC11	6-7	5.873	61.00	[4]
VWA	15-17	6.069	60.98	[5]
SE33	V19-V33	0.717	61.60	[10]
		Combined:	98.99	

it is clear that in most cases, the six systems studied will not give sufficient statistical results. In their paper, the same group of authors [2] emphasize the importance of an appropriate determination of zygosity for clinical studies of twins and for genetic counselling. Regrettably, they did not they did not keep to this requirement in their study. However, we agree with the authors that the six PCR-systems (D1S80, D17S30, ApoB, TC11, VWA and SE33) – *together* with other systems – are well suited for zygosity testing.

One final remark seems worth making: the probabilities of the most frequent and medium-frequent genotypes of the six combined systems are only slightly higher than those which can be achieved by the much cheaper method of questionnaires [11].

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