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Trisomic inheritance

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ABSTRACT

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The theoretical trisomic inheritance is analyzed under certain cytogenetical assumptions. The gametic segregation ratio produced by a duplex individual (*AAa*) is as follows:

$$AA = \frac{1}{6}\left(1 + p.\alpha.\ \frac{1+x}{2}\right);\quad Aa = \frac{2}{6}\left(1 - p.\alpha.\ \frac{1+x}{2}\right);\quad aa = \frac{1}{6}\quad p.\alpha.\quad \frac{1+x}{2};$$

$$A = \frac{2}{6} a = \frac{1}{6}$$

; where 2p is the frequency that one or two crossing-overs take place between the centromere and the locus *A,a*, being *x* the relative frequency of one-crossing-over event. α is the probability that the two centromeres involved in the meiotic paring of two homogous chromosome segments migrate to the same cell pole at anaphase I. The results obtained are compared with those corresponding to the socalled chromosome and chromatid segregations.

INTRODUCTION

Polysomic inheritance is the mode of inheritance which arises when any chromosome (in the case of polysomy or polyploidy) finds more than one partner for chromosome pairing at meiosis (Rieger et al. 1976).

The analysis of tetrasomic inheritance was firstly made under two extreme assumptions, namely, the socalled chromosome segregation (Muller 1914) and chromatid segregation (Haldane 1930). In the first case the gametic segregation ratios result from a meiotic two-by-two distribution of the four homologous chromosomes in which recombination did not occur between the loci analyzed and their respective centromeres. In the second case, gametic segregation ratios are calculated taken only into consideration that each gamete carries two doses of a given locus. Under these assumptions, the gamete frequencies produced by a duplex individual (*AAaa*) are *AA* = 1/6; *Aa* = 4/6, *aa* = 1/6 and *AA* = 3/14, *Aa* = 8/14, *aa* = 3/14, respectively.

A more complete analysis of tetrasomic inheritance was made by Sánchez-Monge (1966) who demonstrated that, under certain cytogenetical assumptions, the gametic segregation ratio produced by a duplex individual (*AAaa*) was: $AA = \frac{1}{6}(1 + 2\,\alpha p)$; $Aa = \frac{4}{6}\,(1 - \alpha\,p)$ $aa = \frac{1}{6}(1 + 2\,\alpha p)$, being 2 p the frequency with which one crossing-over occurs between the locus *A,a* and the centromere, and α the probability that the two centromeres involved in the chromosome segments paired migrate to the same cell pole.

Later, Sánchez-Monge (1972a,b) analyzed in a similar way the gametic segregation and linkage in trisomics.

In this paper, the gametic segregation ratio produced by a duplex trisomic (*AAa*) is calculated based on Sánchez-Monge's model after some modifications.

METHODS

To calculate the gamete frequencies the following assumptions are made:

1. If it occurs, crossing-over takes place in the "centromere-locus *A,a*" chromosome segments without exchange of partners conducing to trivalent formation, there being not any restriction for trivalent meiotic associations involving other chromosome segments. In other words, in the centromere-locus segment crossing-over occurs only between two of the three homologous chromosomes.
2. A 2: 1 segregation of the three homologues occurs at anaphase I
3. The frequency that one or two crossing-overs take place between the centromere and the locus *A,a* is 2p, being *x* and *y* the relative frequencies of one or two crossing-overs, respectively.
4. α is the probability that the two centromeres associated in accordance with the assumption number 1 migrate to the same cell pole at anaphase I.

RESULTS AND DISCUSSION

Three essentially different chromosome dispositions are possible at meiotic prophase, each having a probability of 1/3. According to the model, the gametes produced by each meiotic process depend on the ocurrence of crossing-over (none, one or two events) and the centromere segregation at anaphase I. The gametic segregation ratio can be calculated from Table 1. The total gamete frequencies are: $AA = \frac{1}{6}\,(1 + \text{p}.\alpha.\,\frac{1+\text{x}}{2})$; $Aa = \frac{2}{6}\,(1 - \text{p}.\alpha.\,\frac{1+\text{x}}{2})$ $aa = \frac{1}{6}\,\text{p}.\alpha.\,\frac{1+\text{x}}{2}$; $A = \frac{2}{6}$; $a = \frac{1}{6}$ Obviously, when p = 0 these values coincide with those of the chromosome segregation ($\frac{1}{6}, \frac{2}{6}, \frac{1}{6}, \frac{2}{6}$ and $\frac{1}{6}$, respectively). It is also worth mentioning that when $\alpha = 0$ –that is to say, when the two centromeres associated according to the first assumption of the hypothesis (see Methods) migrate always to different cell poles– the gametic segregation ratio also coincides with that of chromosome segregation, independently of the value of *p*.

On the other hand, when the product $\text{p}.\alpha.\,\frac{1+\text{x}}{2}$ equates to 1/5, the frequencies obtained also applied to the chromatid segregation calculated taking only into account that the gametes having one or two chromosomes (carrying, in consequence, one or two doses of the locus analyzed) are formed in equivalent proportions. In this case, the gametic segregation ratio is: *AA* = 6/30; *Aa* = 8/30; *aa* = 1/30; *A* = 2/6 and *a* = 1/6. It is worthy of mention that the gamete frequencies obtained by Sánchez-Monge (1966) in the case of tetrasomic inheritance also applied to chromatid segregation when $\alpha.p = 1/7$. However, either in the Sánchez-Monge's model for tetrasomic inheritance or in the trisomic inheritance analyzed in this paper, the equations $\alpha.p= 1/7$ or $p.\alpha. \frac{1 + x}{2} = 1/5$ have not any apparent cytogenetical meaning.

The gamete frequencies obtained in this work coincide with those calculated by Sánchez-Monge (1972a) under other cytogenetical assumptions only in the following particular cases: when $p = 0$ or $\alpha = 0$ or when $\alpha = \frac{2x}{1 + x}$. The latter equation has not any apparent cytogenetical meaning, either.

Since the gametic segregation ratio obtained in the present model depends on the value of x, which represents the relative frequency of one-crossing-over events (versus two crossing-overs), it cannot be identified the value p with the genetic distance between the locus A,a and the centromere (measured as the recombination fraction in percent). This is the reason why expressions such as "recombination units", "genetic distances" or equivalents have not been used throughout this paper.

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Table 1.- Gamete formation in a duplex trisomic (*AAa*) according to the meiotic pairing at prophase, the crossing-over events and the centromere segregation. The assumptions of the model are indicated in the text. The total frequencies are calculated by multiplying the probabilities of all the different meiotic events taken into consideration and adding the corresponding values.

Prophase pairing in the centromere-locus chromosomesegment*	Crossing-over events		Anaphase I segregation according to centromere co-orientation		Gamete production				
	Number (0,1,2)	Probability	Configuration	Probability	AA	Aa	aa	A	a
[1-AA 2-AA] 3-aa probability 1/3	any	1	1-AA 3-aa 2-AA	1-α	–	1/2	–	1/2	–
			1-AA 2-AA 3-aa	α	1/2	–	–	–	1/2
	0	1-2p	1-AA 2-AA 3-aa	1-α	1/2	–	–	–	1/2
			1-AA 3-aa 2-AA	α	–	1/2	–	1/2	–
	1	2px	1-Aa 2-AA 3-Aa	1-α	1/4	1/4	–	1/4	1/4
			1-Aa 3-Aa 2-AA	α	1/8	1/4	1/8	1/2	–
1-AA 2-AA 3-aa Probability 1/3		(recip.) 2py1/4	1-AA 2-AA 3-aa	1-α	1/2	–	–	–	1/2
			1-AA 3-aa 2-AA	α	–	1/2	–	1/2	–
	2	(diag.) 2py1/2	1-Aa 2-AA 3-Aa	1-α	1/4	1/4	–	1/4	1/4
			1-Aa 3-Aa 2-AA	α	1/8	1/4	1/8	1/2	–

		(compl.) 2py1/4	1-aa 2-AA 3-AA	$1-\alpha$	–	1/2	–	1/2	–
			1-aa 3-AA 2-AA	α	–	1/2	–	1/2	–
	0	1 - 2p	3-aa 2-AA 1-AA	$1-\alpha$	–	1/2	–	1/2	–
			3-aa 1-AA 2-AA		–	1/2	–	1/2	
	1	2px	3-Aa 2-AA 1-Aa	$1-\alpha$	1/4	1/4	–	1/4	1/4
			3-Aa 1-Aa 2-AA	α	1/8	1/4	1/8	1/2	–
[3-aa / 1-AA] 2-AA probability 1/3		(recip.) 2py1/4	3-aa 2-AA 1-AA	$1-\alpha$	–	1/2	–	1/2	–
			3-aa 1-AA 2-AA	α	–	1/2	–	1/2	–
	2	(diag.) 2py1/2	3-Aa 2-AA 1-Aa	$1-\alpha$	1/4	1/4	–	1/4	1/4
			3-Aa 1-Aa 2-AA	α	1/8	1/4	1/8	1/2	–
		(compl.) 2py1/4	3-AA 2-AA 1-aa	$1-\alpha$	1/2	–	–	–	1/2
			3-AA 1-aa 2-AA	α	–	1/2	–	1/2	–

* Brackets stand for chromosome segments paired. Numbers indicate centromeres. Each letter represents the allele present in the corresponding chromatid.

RESUMEN

En el presente trabajo se analiza la herencia trisómica bajo ciertos supuestos citogenéticos teóricos, obteniéndose las siguientes segregaciones gaméticas producidas por un individuo duplexo (*Aaa*):

$AA = \frac{1}{6}(1 + p.\alpha.\frac{1+x}{2})$; $Aa = \frac{2}{6}(1 - p.\alpha.\frac{1+x}{2})$; $aa = \frac{1}{6}\,p.\alpha.\frac{1+x}{2}$;

$A = \frac{2}{6}\,a = \frac{1}{6}$; donde $2p$ es la frecuencia de que se produzcan 1 ó 2 sobrecruzamientos entre el centrómetro y el locus *A,a*, siendo x la frecuencia relativa con que se produce el suceso de un solo sobrecruzamiento. Por otro lado, α es la probabilidad de que en la anafase I de la meiosis emigren al mismo polo celular los dos centrómetros implicados en el apareamiento meiótico de los segmentos cromosómicos homólogos considerados. Los resultados obtenidos se comparan con los correspondientes a las denominadas segregaciones cromosómica y cromatídica.