An extension of path analysis revisited

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SUMMARY

A method for analysing probabilistic models representing an extension of path analysis, and a model of mixed homogamy based on this method were introduced recently (Rao, Morton & Cloninger, 1979; Cloninger, 1980). However, constraints imposed by the method on linear models, or a rationale for and implications of the mixed homogamy model have not been clearly stated.

A mathematical treatment of the extension of path analysis and of the mixed homogamy model is presented in this paper. Constraints on linear models imposed by the extension are obtained. It is demonstrated that the mixed homogamy model, when applied to analysing nuclear families in a population, implies the following mating system in the population: some individuals choose their mates *strictly* on the basis of group membership, others choose their mates *strictly* on the basis of phenotype, and *no* individual chooses a mate on the basis of *both* group membership and phenotype.

1. INTRODUCTION

Path analysis as a method of analysing probabilistic linear models when some of the variables are unobservable was introduced by Sewall Wright (1921). The method has been applied to different models where 'causal' relationships between analysed variables can be postulated (see recent papers by Cloninger et al. (1983); Karlin et al. (1983) and Wright (1983) for a review, critique and bibliography concerning the use of path analysis in epidemiological genetics). Recently, an extension of path analysis was suggested (Rao, Morton & Cloninger, 1979; Cloninger, 1980) as a device for analysing probabilistic linear models when clear 'causal' relationships between variables cannot be postulated. A model of assortative mating based on this extension of path analysis was called 'mixed homogamy'. The model was criticized by Karlin et al. (1983), who indicated that neither mathematical justification of it nor the constraints imposed by the model on joint distribution of the analysed variables have been discussed. Cloninger et al. (1983) replied to this criticism. However, the question of the constraints on the joint distribution as well as of a specific mating system implied by the mixed homogamy model has remained unanswered.

In this paper, a mathematical investigation of the path analysis extension and of the mixed homogamy model is undertaken. Constraints imposed by this

extension on the joint distribution of the variables in a probabilistic linear model are obtained explicitly. The meaning of the mixed homogamy as a mating system as well as conditions for applicability of the path analysis extension in general are discussed.

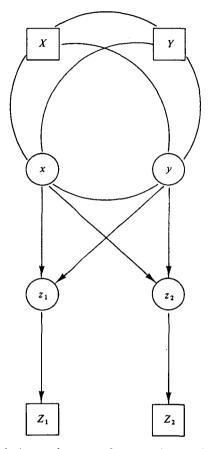


Fig. 1. Nuclear family design under general assortative mating. (X and Y, parental phenotypes; x and y, parental genotypic values; Z_1 and Z_2 , offspring phenotypes; z_1 and z_2 , offspring genotypic values.)

2. GENERAL ASSORTATIVE MATING

Let us consider a nuclear family design under general assortative mating as depicted in Fig. 1, where capital letters are used to denote phenotypes of individuals and lower case letters denote their genotypic values. The lines connecting parental variables are not the paths of path analysis, but simply indicate a correlation between the connected variables. The arrows, on the other hand, are the paths.

We shall assume, as usual in path analysis, that the mean values of all variables are zero, and that the regression of any variable on any subset of variables is linear, e.g. E(x | Y) = a Y, E(Z | X, Y, y) = bX + cY + dy, etc.

We shall also make a number of assumptions simplifying the discussion without restricting its generality.

(a) There is no cultural inheritance of the character under consideration.

(b) The character is sex-independent. This implies that

var(X) = var(Y) = V (phenotypic variance), var(x) = var(y) = v (genotypic variance),

and that the correlation matrix of the parental phenotype–genotype distribution P(X, Y, x, y) is of the form

	X	Y	\boldsymbol{x}	\boldsymbol{y}
X	1	R	b	С
Y	R	1	с	b
x	b	c	1	r
\boldsymbol{y}	С	b	r	1

where R, r, b, c are unknown constants taking values in (-1, 1) and satisfying the restriction that the matrix is positive definite.

(c) The population is at equilibrium with respect to the distribution of the character, thus implying

$$\operatorname{var}(Z_1) = \operatorname{var}(Z_2) = V, \quad \operatorname{var}(z_1) = \operatorname{var}(z_2) = v.$$

(d) The phenotype-on-genotype regressions are

$$E(X|x) = x, \quad E(Y|y = y, \\ E(Z_1|z_1) = z_1, \quad E(Z_2|z_2) = z_2.$$
(1)

It can be shown that, given (1) and the linearity of the genotype-on-phenotype regressions, these regressions are

$$E(x \mid X) = h^{2} X, \qquad E(y \mid Y) = h^{2} Y, \\ E(z_{1} \mid Z_{1}) = h^{2} Z_{1}, \qquad E(z_{2} \mid Z_{2}) = h^{2} Z_{2}, \end{cases}$$
(2)

where $h^2 = v/V$ is the heritability. It follows then that the correlation coefficient b = h, and the correlation matrix of the distribution P(X, Y, x, y) is

Under the above assumptions, the whole linear structure of the nuclear family design is totally specified by the four parameters of the joint parental phenotypegenotype distribution: correlation coefficients R, r, c and heritability h^2 . It is important for the future discussion to notice that there is no constraint under general assortative mating on these parameters; all four of them can be mutually independent (provided only that matrix (3) is a correlation matrix).

The following equations connecting the four parameters of the parental phenotype-genotype distribution with the three observable correlations between

phenotypes of the relatives in nuclear families are easily derived under the above assumptions:

$$\begin{array}{l} \operatorname{corr}\left(\operatorname{parent},\operatorname{parent}\right) = \operatorname{corr}\left(X,\,Y\right) = R,\\ \operatorname{corr}\left(\operatorname{offspr},\operatorname{offspr}\right) = \operatorname{corr}\left(Z_{1},Z_{2}\right) = \frac{1}{2}h^{2}\left(1+r\right),\\ \operatorname{corr}\left(\operatorname{parent},\operatorname{offspr}\right) = \operatorname{corr}\left(X,\,Z\right) = \frac{1}{2}h(c+h). \end{array} \right)$$

$$(4)$$

Notice that these three correlations are the only observable correlations in nuclear families which are independent of each other under the above assumptions a-d. At the same time there are four unknown parmeters. Thus there are fewer equations in the nuclear family design than there are parameters to be estimated. It is important to realize that this unfortunate problem is inherent in the nature of the design and has nothing to do with the specific statistical method used. Whatever statistical method is employed, there must be four unknown parameters but no more than three equations to estimate them, if the linearity of regressions is the only constraint on the joint parental phenotype-genotype distribution. Therefore, if some method of nuclear family analysis operates with a number of independent parameters less than four, it must be that some assumptions, albeit implicit, have been made imposing additional constraints on the distribution P(X, y, x, y) besides the linearity of regressions.

When path analysis is applied to nuclear families under 'pure' social or 'pure' phenotypic homogamy, assumptions reducing the number of independent parameters in parental phenotype–genotype distribution are implied by the 'causal' relationships between variables resulting from explicit definitions of these homogamies as specific mating systems.

3. SOCIAL HOMOGAMY

The following definition was given to this mating system: 'Under social homogamy mates choose each other on the basis of their group membership, which generates primary correlations between the genotypes and environments of spouses, and the phenotypic correlation between spouses becomes secondary' (Rao & Morton, 1980). In other words (neglecting for simplicity the primary environmental correlation), social homogamy is a system of assortive mating under which the probability that two individuals mate is completely determined by their genotypes, independently of their phenotypes, i.e.

$$P(\text{mate} \mid X, Y, x, y) = P(\text{mate} \mid x, y).$$
(5)

It is not difficult to prove that this is equivalent to the 'causal' relationship between genotypes and phenotypes in parents:

$$P_A(X | Y, x, y) = P_A(X | x),$$

$$P_A(Y | X, x, y) = P_A(Y | y)$$
(6)

(subscript A indicates social homogamy). That is, the probability for a parent to be of a particular phenotype is completely determined under social homogamy by the parent's genotype. This 'causal' genotype-phenotype relationship constitutes the rationale for the path diagram of social homogamy (Fig. 2a).

It can be shown that (6) leads to the following dependency of the correlation coefficients R_A and c_A on the rest of the correlation matrix (3) for the distribution $P_A(X, Y, x, y)$:

$$R_A = r_A h_A^2, \quad c_A = r_A h_A. \tag{7}$$

Thus, the number of independent parameters in the nuclear family design is reduced in the case of social homogamy from four to two: r_A and h_A^2 , and the three equations to estimate them follow from (4):

$$\begin{array}{l} \operatorname{corr}(X, Y) = r_A h_A^2, \\ \operatorname{corr}(Z_1, Z_2) = \frac{1}{2} h_A^2 (1 + r_A), \\ \operatorname{corr}(X, Z) = \frac{1}{2} h_A^2 (1 + r_A). \end{array} \right\}$$
(8)

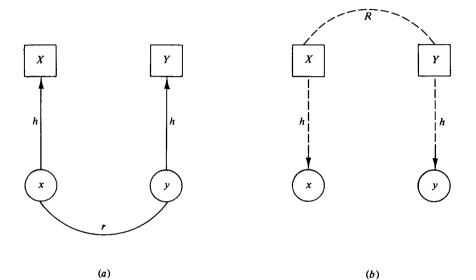


Fig. 2. Path diagrams of the joint parental distributions under 'pure' homogamies: (a) social homogamy; (b) phenotypic homogamy (r, genotypic correlation; R, phenotypic correlation; h^2 , heritability).

The same system of equations is obtained if the rules of path analysis are applied to the diagram in Fig. 2a. Since there are more equations then parameters being estimated, one of the equations is usually used to test goodness of fit of the model.

4. PHENOTYPIC HOMOGAMY

The following definition was given to this mating system: 'Under this system, mates choose one another strictly on the basis of their phenotypes' (Rao & Morton, 1980), i.e.

$$P(\text{mate} \mid X, Y, x, y) = P(\text{mate} \mid X, Y).$$
(9)

This can be proved to imply the 'causal' phenotype genotype relationship in parents under phenotypic homogamy:

$$P_{B}(x|X, Y, y) = P_{B}(x|X), \quad P_{B}(y|X, Y, x) = P_{B}(y|Y), \quad (10)$$

(subscript B indicates phenotypic homogamy). In other words, the probability for a parent to be of a particular genotype is completely determined by the parent's phenotype. This 'causal' relationship constitutes the rationale for the path diagram of phenotypic homogamy (Fig. 2b).

It can be shown that (10) leads to the following dependency of correlation coefficients r_B and c_B on the rest of the correlation matrix (3) for the distribution $P_B(X, Y, x, y)$:

$$r_B = R_B h_B^2 \quad c_B = R_B h_B. \tag{11}$$

Thus the number of independent parameters in the nuclear family design is reduced under phenotypic homogamy from four to two, R_B and c_B , and there are three equations to estimate them:

$$\begin{array}{c} \operatorname{corr} (X, Y) = R_B, \\ \operatorname{corr} (Z_1, Z_2) = \frac{1}{2} (1 + R_B h_B^2) h_B^2, \\ \operatorname{corr} (XZ) = \frac{1}{2} (1 + R_B) h_B^2. \end{array} \right\}$$
(12)

Again there are more equations than parameters being estimated, and one of the equations is usually used for testing goodness of fit of the model.

5. MIXED HOMOGAMY

It should be emphasized that the reduction in the number of independent parameters (and, hence, the applicability of path analysis) in the nuclear family design under pure social and phenotypic homogamies is due to the constraints on the parental phenotype-genotype distribution ('causal' relationships) implied by the *explicit definitions* of these homogamies as specific mating systems.

Unlike social and phenotypic homogamies, mixed homogamy introduced by Rao, Morton & Cloninger (1979) has never been explicitly defined as a mating system. It was simply stated that 'a generalization incorporating phenotypic and social homogamy as two special cases is called mixed homogamy' (Rao & Morton, 1980), and the model was introduced as a 'superposition' of the path diagrams for social and phenotypic homogamies (Fig. 3) together with a set of rules for manipulating the superposed diagram: parallel paths (Rao & Morton, 1980), or co-paths (Cloninger, 1980). However, no mathematical justification of the path diagram superposition and of the rules for manipulating the superposed diagrams has been presented. The meaning of parameters in the superposed diagram (Fig. 3) is also unclear. It is said that p is the 'primary correlation between parental phenotypes, not due to secondary resemblance through social homogamy', and mis the 'correlation between parental genotypes through social homogamy' (Rao et al. 1979). Does this mean that p and m are respectively the correlation between phenotypes of parents under *pure* phenotypic homogamy and the correlation between genotypes of parents under *pure* social homogamy? If not, then what are they?

This lack of mathematical justification inevitably invited the question: 'How do we know that such superposed models do not involve contradictions in their basic structure or nonrealizable constraints on parameters?' (Karlin *et al.* 1983). Indeed, consider a situation when observable correlations in nuclear families take the following values (Rao *et al.* 1982):

$$\begin{array}{c} \operatorname{corr} (X, Y) = 0.513, \\ \operatorname{corr} (Z_1, Z_2) = 0.513, \\ \operatorname{corr} (X, Z) = 0.485. \end{array} \right)$$
(13)

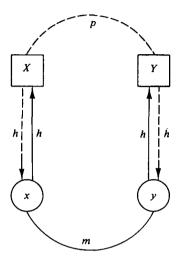


Fig. 3. Path diagram of the joint parental distribution under the 'mixed homogamy' model (h^2 heritability; p and m, parameters of the 'superposed' diagram).

If the rules of manipulating the superposed diagrams (Rao and Morton, 1980; Cloninger, 1980) are applied, the following relationships between the observable correlations and parameters of the mixed homogamy diagram (Fig. 3) emerge:

$$corr (X, Y) = p + mh^{2},corr (Z_{1}, Z_{2}) = \frac{1}{2}(1 + m + ph^{2})h^{2},corr (X, Z) = \frac{1}{2}(1 + m + p)h^{2}.$$
(14)

The substitution of the values of observable correlations from (13) into (14) yields after some transformations the following quadratic equation for the heritability:

$$(h^2)^2 - 0.457h^2 + 0.0560 = 0.$$
⁽¹⁵⁾

This equation has no real solution. Thus, the mixed homogamy model (Fig. 3) cannot resolve the situation when observable correlations are as in (13). Some people would argue that this is a result of neglected environmental correlations, and that if these correlations were incorporated, the situation would have been resolved by the mixed homogamy model. This indeed may very well be true. The important point, however, is that this could just as well be not true, since the situation can easily be resolved even with zero environmental correlations under general assortative mating. For example, if the heritability

$$h^2 = 0.64$$

and other parameters of the joint parental distribution P(X, y, x, y) are: R = 0.513, r = 0.603, c = 0.412 (matrix (3) in this case is positive definite), the observable correlations will take values as in (13). This is not the only joint parental distribution resolving the situation. The set of parameters including the heritability

$$h^2 = 0.52$$

and R = 0.513, r = 0.973, c = 0.624 also will do. As a matter of fact there is, of course, a continuum of joint parental distributions resulting in observable correlations as in (13).

The problem posed by the above example becomes even more acute when the mixed homogamy model can resolve observable correlations and does provide estimates of some genetic and other parameters in a population. Where is the guarantee that the model is applicable to this population and, therefore, that the estimates of the parameters are indeed correct? In the case of a pure homogamy model, its applicability to a particular population is easily decided on the basis of whether it is believed or not that the corresponding explicitly defined mating system does operate in the population. A similar decision about the applicability of the mixed homogamy model cannot, however, be made, since the corresponding mating system has not been defined.

The following relationships between parameters of the joint parental distribution P(X, Y, x, y) and parameters of the mixed homogamy model are obtained if the rules of manipulating superposed diagrams are applied to the diagram in Fig. 3:

$$R = p + mh^2, \quad r = m + ph^2, \quad c = (m + p)h.$$
 (16)

Recall that under general assortative mating, parameters R, r, c and h^2 can be mutually independent (provided matrix (3) is a correlation matrix). Given (16), however, this is not true any more, and the following relationship must hold for these parameters in order for the mixed homogamy model to be applicable:

$$\frac{c}{R+r} = \frac{1}{1+h^2}.$$
 (17)

Notice that this relationship does hold for the parameters of the joint parental distribution in the case of a pure homogamy. Indeed, (17) follows directly from either (7) or (11), which in turn result from the 'causal' relationships under the pure homogamies. Since these 'causal' relationships are implied by the definitions of these homogamies as specific mating systems, relationship (17) is also *implied* in the case of pure homogamies by their definitions and need not be *imposed* in order for the model to be applicable. The applicability of pure homogamy models, as has already been mentioned, is decided on the basis of their definitions as specific mating systems and not on the basis of whether relationship (17) holds or not.

In the case of the mixed homogamy model, though, since there is no definition of this homogamy as a specific mating system, relation (17) is not implied, but rather represents a constraint that must be *imposed* on the joint parental distribution in order for the model to be applicable. The applicability of the model to a particular population should, therefore, be decided on the basis of whether constraint (17) is satisfied in this population or not. Notice, however, that this cannot be tested, since (17) involves unobservable parameters. In order to find out what mating systems are implied by the mixed homogamy model, i.e. under what mating systems, besides the pure homogamies, constraint (17) is satisfied, let us consider the following decomposition of the joint parental distribution under general assortative mating:

$$P(X, Y, x, y) = f_A P_A(X, y, x, y) + f_B P_B(X, Y, x, y) + f_{AB} P_{AB}(X, Y, x, y), \quad (18)$$

where f_A is the proportion of individuals choosing their mates *strictly* on the basis of the mate's group membership (pure social homogamy), f_B is the proportion of individuals choosing mates *strictly* on the basis of the mate's phenotype (pure phenotypic homogamy) and f_{AB} is the proportion of individuals choosing mates on the basis of *both* the mate's group membership and phenotype (intersection of the pure homogamies). $P_A(X, Y, x, y)$, $P_B(X, Y, x, y)$ and $P_{AB}(X, Y, x, y)$ are the joint parental distributions under pure social, pure phenotypic and the intersection of both homogamies, respectively. The decomposition (18) is, of course, always possible, provided there are no other mating systems operating in the population, besides the three mentioned. Given the linearity of all regressions, (18) yields the following relationship for the elements of the correlation matrix (3):

$$R = f_A R_A \frac{V_A}{V} + f_B R_B \frac{V_B}{V} + f_{AB} R_{AB} \frac{V_{AB}}{V},$$

$$r = f_A r_A \frac{v_A}{v} + f_B r_B \frac{v_B}{v} + f_{AB} r_{AB} \frac{v_{AB}}{v},$$

$$c = f_A c_A \sqrt{\frac{v_A V_A}{v V}} + f_B c_B \sqrt{\frac{v_B V_B}{v V}} + f_{AB} c_{AB} \sqrt{\frac{v_{AB} V_{AB}}{v V}},$$

$$h = f_A h_A \sqrt{\frac{v_A V_A}{v V}} + f_B c_B \sqrt{\frac{v_B V_B}{v V}} + f_{AB} c_{AB} \sqrt{\frac{v_{AB} V_{AB}}{v V}}.$$
(19)

(Indices A, B and AB indicate variables related to social, phenotypic and the intersection of both homogamies, respectively, whereas unindexed variables relate to general assortative mating.) It should be be remembered that, while there are restrictions on the parameters of distributions $P_A(X, Y, x, y)$ and $P_B(X, Y, x, y)$ implied by the 'causal' relationships of the pure homogamies, there is no restriction on the parameters of the distribution $P_{AB}(X, Y, x, y)$; under general assortative mating they can take any values, provided matrix (3) is a correlation matrix.

Since there are more independent variables in (19) than even the original four, it is obvious that assumptions reducing their number need to be made. We shall make the following imoportant assumption.

(i) The decision by an individual whether to choose a mate on the basis of the mate's phenotype or on the basis of the mate's group membership or on the basis of both does not depend on the individual's own phenotype or group membership. This is equivalent to assuming that the genotypic and phenotypic distributions are the same among individuals mating through social homogamy as among those mating through phenotypic homogamy or through the intersection of both. This implies

$$\begin{array}{l}
V_{A} = V_{B} = V_{AB} = V, \\
v_{A} = v_{B} = v_{AB} = v, \\
h_{A}^{2} = h_{B}^{2} = h_{AB}^{2} = h^{2}.
\end{array}$$
(20)

With this assumption and taking into account (7) and (11), system (19) is reduced to

$$\begin{array}{l} R = f_{A}r_{A}h^{2} + f_{B}R_{B} + f_{AB}R_{AB}, \\ r = f_{A}r_{A} + f_{B}R_{B}h^{2} + f_{AB}r_{AB}, \\ c = (f_{A}r_{A} + f_{B}R_{B})h + f_{AB}c_{AB}. \end{array}$$

$$(21)$$

Given this, constraint (17) implies

$$f_{AB}[(R_{AB} + r_{AB})h - c_{AB}(1 + h^2)] = 0.$$
⁽²²⁾

In order for (22) to be satisfied, it is necessary that either

$$f_{AB} = 0, \tag{23}$$

or

$$\frac{c_{AB}}{R_{AB} + r_{AB}} = \frac{h}{1 + h^2},$$
(24)

i.e. either there is no individual mating through the intersection of social and phenotypic homogamies in the population or, if the population does have such individuals, parameters of the joint parental distribution among them must satisfy a relationship analogous to the constraint (17), which is imposed on the joint parental distribution in the whole population in order for the mixed homogamy model to be applicable.

Considering relationship (24), notice that, although a similar relationship is implied in the case of pure homogamies by their 'causal' structures, in the case of the intersection of the two homogamies relationship (24) is not implied, since no 'causal' structure can be associated with this mating system. In a particular population parameters R_{AB} , r_{AB} , c_{AB} and h^2 can take arbitrary values (provided only that matrix (3) is a correlation matrix). To expect that relationship (24) will happen to hold in a particular population is equivalent, therefore, to expecting that four continuously distributed random variables will happen to form a specific combination. The probability of such an event is, of course, infinitesimal. Relationship (24) will, therefore, almost certainly never hold in a particular population and it can be safely ignored in the context of the applicability of the mixed homogamy model.

Thus condition (23), or the absence or individuals mating through the intersection of social and phenotypic homogamies, represents the only realistic implication of constraint (17) and, hence, of the mixed homogamy model.

Let us now analyse the meaning of the parameters of the superposed diagram (Fig. 3), given that the mixed homogamy model is applicable, i.e. (23) is satisfied. Given this, (21) yields

$$\left. \begin{array}{l} R = f_A r_A h^2 + f_B R_B, \\ r = f_A r_A + f_B r_B h^2, \\ c = (f_A r_A + f_B r_B) h. \end{array} \right\}$$

$$(25)$$

Comparing this with (16), it may be concluded that

$$p = f_B R_B, \quad m = f_A r_A. \tag{26}$$

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Thus parameters of the superposed diagram (Fig. 3) have the following meaning (when the mixed homogamy model is applicable): p is the phenotypic correlation of mates under pure phenotypic homogamy weighted by the proportion of individuals who mate through this mating system; m is the genotypic correlation of mates under pure social homogamy weighted by the proportion of individuals who mate through this mating system.

6. CONCLUSION

The mixed homogamy model is applicable to analysing nuclear families in a population only if the following can be assumed.

(i) An individual in the population chooses a mate either *strictly* on the basis of the mate's group membership, or *strictly* on the basis of the mate's phenotype, and *no* individual chooses a mate on the basis of *both* the mate's group membership and phenotype.

(ii) The decision by an individual whether to choose a mate on the basis of the mate's group membership or on the basis of the mate's phenotype does not depend on the individual's own group membership or phenotype.

More generally, the extension of path analysis based on the superposition of path diagrams is applicable to a probabilistic linear model, only if the model is a mixture of *mutually exclusive* causal structures, and the marginal distributions of variables are the same in all of the causal structures.

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