

Application of Markov models for analysis of development of psychological characteristics

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ABSTRACT

A technique to study combined influence of environmental and genetic factors on the base of changes in phenotype distributions is presented. Histograms are exploited as base analyzed characteristics. A continuous time, discrete state Markov process with piecewise constant interstate transition rates is associated with evolution of each histogram. The technique was applied to IQ longitudinal data (6- and 14-year children) and made it possible to draw conclusions concerning development of Russian children before school and during the first stage of school education as well as dependence of environmental and genetic influence on IQ level.

INTRODUCTION

Combination of genetic and environment factors is a logical consequence of the grouping humans into population of self-determining individuals who share both genes and environment in common. Combined genotype-environment effects conditioned by assortative mating, genotype-environment correlation, genotype-environment interaction, etc. are referred to here, as a whole, as the results of influence of some *systematic factor*. Longitudinal analysis of such influences is frequently of interest in psychological investigations.

Means for this analysis are usually include various tests for concordance, factor analysis of variance/covariance structures and, as one of its most advance variants, analysis of simplex models (Guttman, 1954; Boosma, Martin & Molenaar, 1989; Dolan, Molenaar & Boosma, 1991). Nevertheless, analyzing such integrating characteristic as variances, covariances and means, a researcher loses a lot of information. As a rule, he also has difficulties during extrapolation of obtained results to the time domains between observation points and during study of dependences of factor effects on different levels of individual characteristics. That is why the search of other solution techniques based on new principles is topical.

Presented here is the technique intended for studying evolution of measured phenotype distributions with age. It follows the ideas stated in our previous paper (Kuravsky & Malykh, 2000) where genetic differences between twins and effects of non-shared environment were estimated. As earlier, histograms are used as base analyzed characteristics. However now they describe individual characteristic distributions for unrelated individuals, but not the differences in twin pairs. Using measurements in a series of time points, the technique discussed makes it possible to determine how the systematic factor changes an initial phenotype distribution during the selected time period.

The technique is demonstrated on the analysis of IQ longitudinal data (6- and 14-year children). The main purpose of this work was to investigate combined environmental and genetic influences on population as functions of age and IQ level. Some interesting conclusions concerning development of Russian children before school and during the first stage of education were drawn from the analysis. In particular, it was shown that standard education promoted development of unable individuals more effectively than development of capable ones. Results of analysis on the base of the presented approach were compared with the output obtained with the aid of classical phenotypic simplex model. Finally, a so-called 2-dimensional model to compare combined genotype-environment effects leading to changes in population phenotype and effects of non-shared environment evaluated on twin data is discussed.

Previous research covering similar areas include Markov chain Monte Carlo methods (Eaves & Erkanli, 2003; Creutz, 1979; Gelfand & Smith, 1990) and random utility models (Falmagne & Regenwetter, 1996). Both approaches carry out histogram estimation. The first one constructs a Markov chain on the parameter space of unknown quantities such that, starting with a series of trial values (e. g. means, regressions, etc.), after an initial series of iterations, successive iterations represent samples from the unknown joint stationary distribution. The Gibbs sampler (Creutz, 1979; Gelfand & Smith, 1990) is popular approach of this type to construct a Markov chain with desired properties. As distinct from the approach presented in this paper, Markov chain Monte Carlo methods are not acceptable for longitudinal analysis of observed data because of both their inefficient representation of evolutionary processes and inability to solve the inverse problem, viz.: to derive necessary development characteristics from the observed distributions.

The second approach is represented by a model for approval voting. It is based on the notion that each voter has a personal ranking of the alternatives, with a random utilities being defined by the probability distribution on the set of all rankings. However, this method is overly attached to the details of a particular application. Its model is static and, in contrast to the technique presented in this paper, is not acceptable for representation of evolutionary processes. Moreover, the random utility model fails in case of violations of the basic axiom system (the authors showed a corresponding example).

METHOD

In the model presented, the distribution of individual characteristics at the moment of birth describes influences of heritability and perinatal/prenatal environmental factors. As a result of subsequent influences of the systematic factor, initial distributions are transformed to current ones during the time of observation. This idealization is based on the results of twin studies of IQ, which demonstrated that both environment and genetic influences on individual differences are present and dynamic throughout development (Eaves, Eysenck & Martin, 1989; Malykh, Egorova & Meshkova, 1998). As genetically uninformative samples are analyzed, effects of these factors are not separated here.

It is important for the parameter of interest to be measured in a time-independent scale (i. e. the testing technique must take into account the natural age development as is the case for IQ).

A continuous time discrete state Markov process with piece-wise constant interstate transition rates¹ is associated with evolution of each histogram. Given a continuous trait, the available actual range of the analyzed quantity is divided into several intervals. These intervals are considered as separate discrete states in which the trait value has some probability to find itself. In due course transitions between the states are the case. The number of these states is determined by desirable precision of estimates and available sample size².

¹ These processes are frequently used to solve problems of the queuing theory.

² Loss of information due to discretization is not a serious problem, especially if large enough samples are possible such that state intervals are smaller than measurement error.

Some considerations concerning variation of state interval length are presented in our previous paper (Kuravsky & Malykh, 2003).

This model may be represented by a graph (Figure 1) in which nodes (depicted as rectangles) correspond to the states; branches (depicted as arrows) correspond to transitions. The trait evolution process may be imagined as a random walk along the graph from one state to another following the arrows. Time is supposed to be continuous. State-to-state transitions are instantaneous and take place at random time points. These transitions are effects of the systematic factor. To describe it mathematically means to show how this factor has an influence on population.

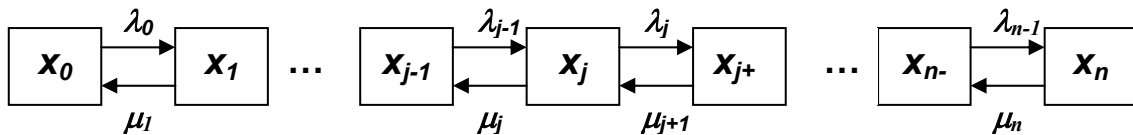


Figure 1. The model used to describe evolution of phenotype distributions with age, where the x_j ($j = 0, 1, \dots, n$) are states, the λ_p ($p = 0, 1, \dots, n - 1$) and μ_q ($q = 1, 2, \dots, n$) are flow rates.

It is assumed that state-to-state transitions (corresponding to each branch of the graph) meet the properties of Poisson's flows of events (Ovcharov, 1969). It may be proved (Ovcharov, 1969) that the number of events X in these flows falling into any interval of the length τ adjoining to time point t is described by the Poisson distribution:

$$P_{t,\tau}(X = m) = \frac{(a(t,\tau))^m}{m!} e^{-a(t,\tau)},$$

where $P_{t,\tau}(X = m)$ is the probability of appearance of m events during the considered interval, $a(t,\tau)$ - mean number of events falling into an interval of the length τ adjoining to time point t . Only stationary flows (where $a(t,\tau) = \eta\tau$, $\eta = \text{const}$) will be taken up here. Parameter η is the rate of a stationary flow. It is equal to mean number of events per unit time interval. Mean time interval between two adjacent events in this flow is $1/\eta$.

The aforementioned assumptions concerning the nature of event flows are usual for applications as these flows (or flows which close to them) are frequently take place in reality because of limit theorems for events (Ovcharov, 1969).

The system shown in Figure 1 is a finite chain of $n + 1$ states where transitions from the state x_j ($j = 1, 2, \dots, n - 1$) are possible only to the preceding state x_{j-1} or to the next state x_{j+1} . Available from the states x_0 and x_n are the states x_1 and x_{n-1} correspondingly.

Flow rates are denoted as λ_p and μ_q ($p = 0, 1, \dots, n - 1$ and $q = 1, 2, \dots, n$). Parameters λ_p define how the influences of the systematic factor under study promote increase of the examined phenotype values. Parameters μ_q describe the inverse effect of this factor – decrease of the phenotype.

If one denotes possible bottom and top limits of parameter change as D_{bot} and D_{top} , the state x_0 corresponds to the interval from D_{bot} to $D_{bot} + \frac{D_{top} - D_{bot}}{n+1}$, the state x_1 - to the interval from $D_{bot} + \frac{D_{top} - D_{bot}}{n+1}$ to $D_{bot} + 2\frac{D_{top} - D_{bot}}{n+1}$, and so on. The following set of ordinary differential equations (Ocharov, 1969) may be drawn to describe the time history of state probabilities:

$$\frac{dp_0(t)}{dt} = -\lambda_0 p_0(t) + \mu_1 p_1(t);$$

.....

$$\frac{dp_j(t)}{dt} = -(\lambda_j + \mu_j)p_j(t) + \lambda_{j-1}p_{j-1}(t) + \mu_{j+1}p_{j+1}(t);$$

.....

$$\frac{dp_n(t)}{dt} = -\mu_n p_n(t) + \lambda_{n-1} p_{n-1}(t)$$

where $p_k(t)$ is the probability to be within the state x_k at the time point t ($k = 0, 1, \dots, n$).

To integrate these equations, one has to assign initial conditions

$$p_0(0), p_1(0), \dots, p_n(0); \sum_{k=0}^n p_k(0) = 1.$$

The normalization condition $\sum_{k=0}^n p_k(t) = 1$ is valid at any time point. It is postulated that

$t = 0$ is the moment of birth.

Since the quantities that are formed as a result of influence of many different elementary factors (such as IQ) may be considered as asymptotically normal ones, it is supposed that at the moment of birth and other test time points of interest, these characteristics may be approximately described by some normal distributions. Mean m_0 and standard deviation σ_0 at point $t = 0$ characterize the initial distribution.

So, estimation of systematic effects are brought to the calculation of mean m_0 , standard deviation σ_0 and flow rates λ_p and μ_q . Values ensuring the best fit of expected and observed frequencies of falling into each state at the specified time points are taken as estimations of these free parameters. Estimated values are considered as the characteristics of systematic effects which have become apparent during observations. Normality of phenotype distributions at the moments of observations (as is the case for IQ study) is actually an additional constraint for selection of flow rates.

In case of normally distributed phenotypes, free model parameters are to be determined as functions of means and standard deviations of corresponding normal distributions to avoid model adjustment to disturbances induced by sampling errors. Such smoothing of observed histograms reduces the dependence of final conclusions from these errors.

Expected state probabilities are calculated by means of integration of the presented set of differential equations. Expected frequency to fall at the k -th state equals to $p_k N$, where p_k – probability of being in this state, N – number of cases. Corresponding original observed frequencies F_k result from measurements obtained during longitudinal study.

Provided that the given expected frequencies describe observed data³, the following Pearson statistic is distributed asymptotically according to a chi-square distribution (Cramer, 1946):

$$\sum_{k=0}^n \frac{(F_k - p_k N)^2}{p_k N}.$$

One should regard this sum as a goodness-of-fit measure in the sense that its large values correspond to bad fit and its small values correspond to good fit. Under certain general conditions (Cramer, 1946; Fisher, 1924), the number of degrees of freedom is equal to $n - l$, where l is the total number of free parameters determined from the sample under study, and serves as a standard by which one can judge whether such measure is large or small.

Estimations of free parameters are found as the values minimizing the sum of goodness-of-fit measures at specified time points in which observed data are available, with the original random observed frequencies in the expressions of Pearson statistic being replaced by the corresponding frequencies of the best-fitted normal distributions. Such substitutions disable the opportunity to use a chi-square distribution in the test for concordance. However, one can use sum of the aforementioned statistics simply as a criterion for minimization to find the best-fitted values⁴ and, then, carry out the test for concordance of the obtained model and original observed data.

The employed estimation procedure consists of *three stages*. *On the first stage*, some numerical integration scheme for the aforementioned differential equations is coded to calculate all $p_k(t)$ using the *Microsoft® Excel* spreadsheet. The probability functions are computed with some specified time step h from initial zero time point to the given specified upper time bound. Runge-Kutta methods (Bakhvalov, 1975) (or their equivalents) proved to be sufficient to get acceptable accuracy of solution. It is of vital importance that *Excel* supports dynamic links between cell contents, viz.: if one locates current values of free parameters and time step h in the separate cells to which the cells containing formulas for calculation of $p_k(t)$ and initial state probabilities are referred, all the solution will be automatically modified when values of free parameters are changed.

On the second stage, parameters of best-fitted normal distributions for observed frequencies F_k are estimated, and, later, the frequencies defined by these distributions are used instead of the observed ones. Parameters of the best-fitted distributions may be calculated either as sample point estimates or by the chi-square minimum approach directly.

On the third stage, a numerical optimization procedure to get required values of free parameters is run. The authors used a macros realizing the procedure of non-linear optimization called *Generalized Reduced Gradient (GRG2)*.

It is necessary to note that this technique based on capabilities of spreadsheets, in fact, determines coefficients of model differential equations using a given observed solution (the inverse problem is solved).

³ Null hypotheses.

⁴ In fact, we apply the least-squares method instead of the method of chi-square minimum in its classical form. The expression of Pearson statistic is used formally as normalized least squares criterion.

RESULTS

In the following sections, application of the presented approach to analysis of IQ longitudinal data is discussed. Test data for 94 six-year old children and 70 fourteen-year old children are analyzed. All the children entered standard Russian schools. Measurements of general IQ (GIQ) were carried out twice: at age 6 and age 14 (for the same children). Expected frequencies were adjusted to observed ones at the time points corresponding to the 6- and 14-year age. IQ range from 60 to 160 was taken into account. The model to be fitted was represented by a chain containing 20 states.

Chi-square tests show no significant differences between observed frequencies for selected IQ intervals and corresponding expected frequencies of normal distributions ($p = 0.83$ for 6-year sample and $p = 0.88$ for 14-year sample). Therefore the hypothesis of normality fits our data.

According to F -test, there are no significant differences between variances at ages 6 and 14 ($p = 0.12$). So we can accept the hypothesis of homogeneity of variances in the following considerations and use their common value in estimations. In its turn, t -test shows highly significant differences in means between the samples in question ($p < 0.00001$).

The sum of goodness-of-fit measures at the 6- and 14-year time points was minimized by selecting the following free parameters:

- flow rates λ_p and μ_q for the range 0-6 years;
- flow rates λ_p and μ_q for the range 6-14 years;
- mean m_0 and standard deviation σ_0 .

To keep reasonable balance between the model complexity and depth of investigation, we have to use minimal number of free parameters that ensures acceptable model fitting to observed data and capabilities for analysis of interest. To determine this number, three models of different capacity were compared. In *the first model*, flow rates were assumed to be the same for the ranges 60-85, 85-110, 110-135, 135-160 of IQ units: $\lambda_0 = \lambda_1 = \lambda_2 = \lambda_3, \dots, \lambda_{16} = \lambda_{17} = \lambda_{18} = \lambda_{19}$; $\mu_1 = \mu_2 = \mu_3 = \mu_4, \dots, \mu_{17} = \mu_{18} = \mu_{19} = \mu_{20}$. (It is reasonable to assume that flow rates do not have great changes with GIQ growth.) In *the second model*, these IQ ranges are wider: 60-110 and 110-160. And in *the third model* flow rates do not depend on IQ values. Results of model fitting are given in Table 1.

Table 1. Fitting models of difference capacity.

Model No	Goodness-of-fit measure (with respect to the frequencies of best-fitted normal distributions)	Chi-square goodness measure (with respect to the original observed data)	Degrees of freedom	p -value
1	0.43	19.07	8	0.01
2	0.79	18.75	16	0.28
3	1.75	18.37	20	0.56

One should select the best-fitted model in the sense of our goodness-of-fit measure, which ensures the acceptable correspondence to observed data. Since *the first model* did not agree with them ($p=0.01$), *the second model* ($p=0.28$) was selected for the following analysis. The *Third model* was rejected as it had the worse value of the minimization criterion.

In calculations, one year was used as a unit of time measurement. The modified Euler method was coded in the spreadsheet as a numerical integration scheme (with time step from 0.006 to 0.008 years).

Figure 2 and Table 2 present obtained estimates of flow rates and IQ initial mean and standard deviation. Observed and expected state probabilities for ages 6 and 14 are presented in Figure 3. Given in Figure 4 are plots showing the envelopes of distributions of expected state probabilities at the moment of birth and test time points ($t = 6, 14$). IQ of neonates is considered here as an extrapolated characteristic describing their later abilities. It cannot be estimated directly.

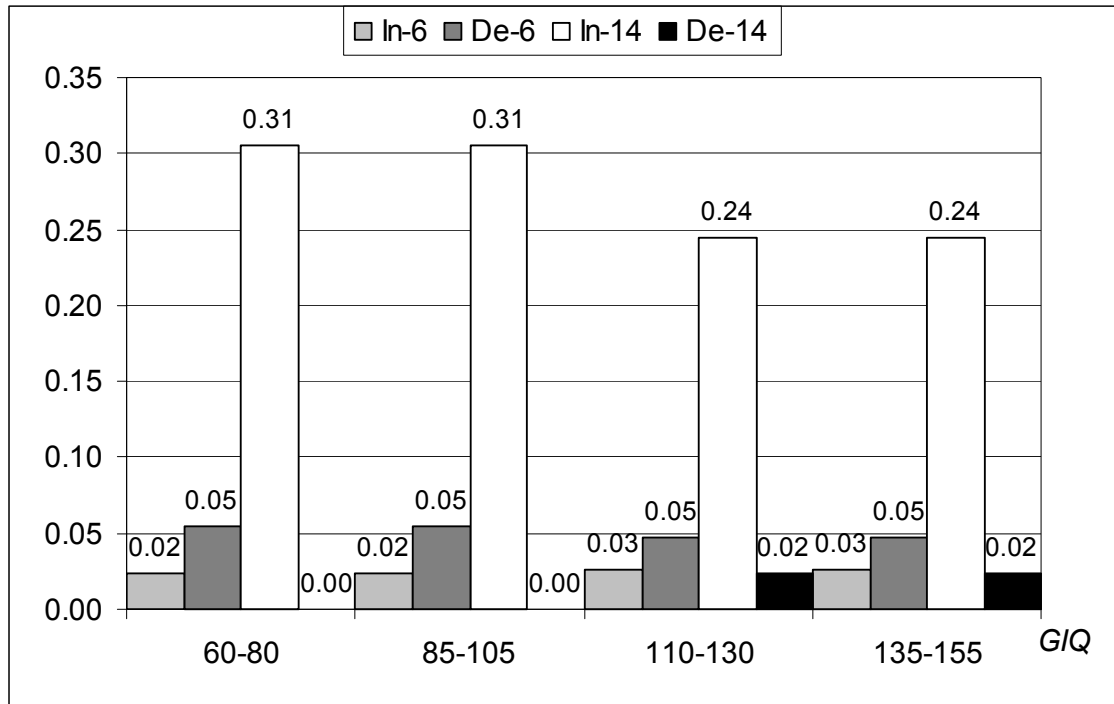


Figure 2. Estimates of flow rates (*In* – increase; *De* – decrease).

Table 2. Model 2: parameters of expected normal distributions.

Age	Mean of normal distribution	Standard deviation of normal distribution
0 years	99.18	11.81
6 years	98.24	12.63
14 years	109.03	12.63

To judge the correctness of model application, it is necessary to find out how stable are the estimations of free parameters as well as derived conclusions to variations of initial data. As to the given problem, such three independent statistics, as sample means for ages 6 and 14 and the respective common sample standard deviation, together with the important assumption on normality of distributions, determine our model. A set of solutions corresponding to prescribed initial data confidence intervals was analyzed in paper^[15] to substantiate stability of the obtained results.

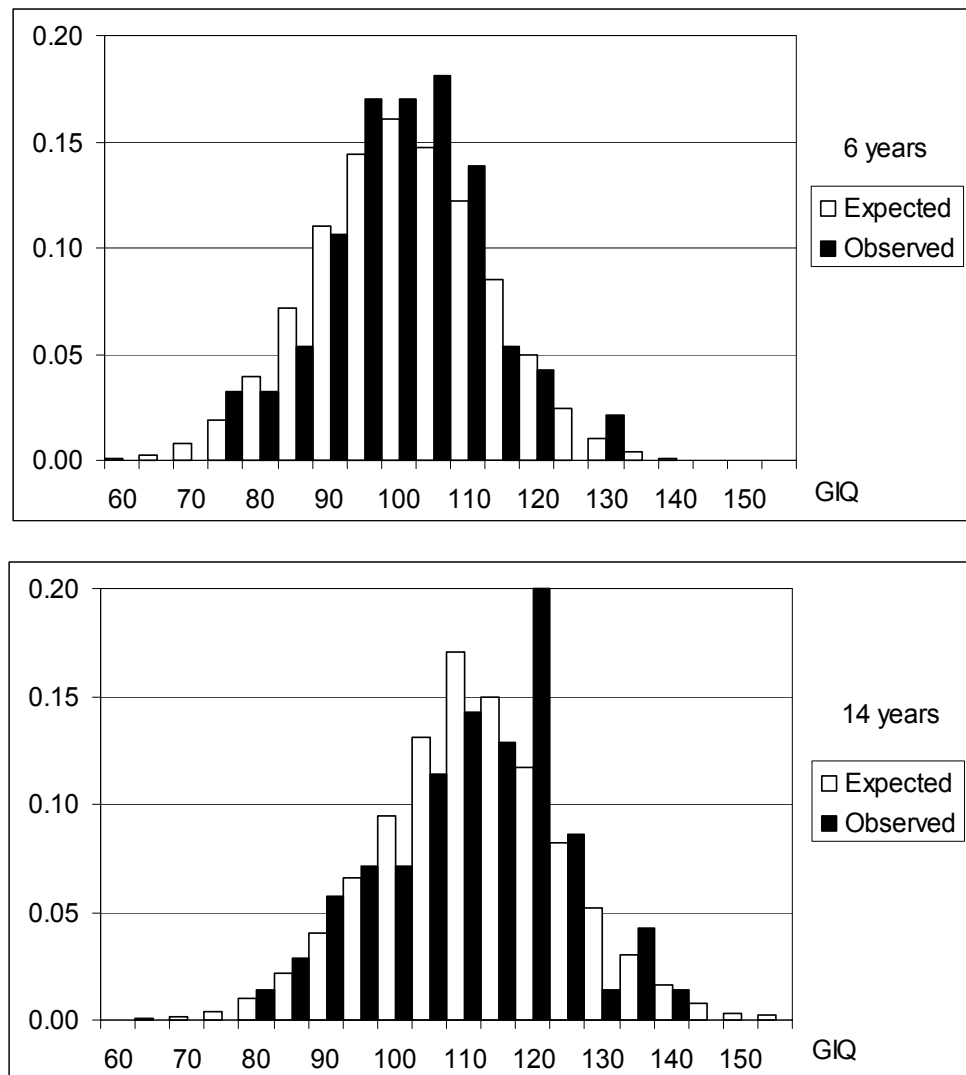


Figure 3. Observed and expected state probabilities for ages 6 and 14.

DISCUSSION

As a rule, Russian children start their school education at age 6 or 7. Therefore the range 0-6 corresponds mainly to preschool development, and the range 7-14 - to the first stage of school development. Taking this fact into account, the following qualitative conclusions, which are valid within 80% confidence region of the above mentioned independent statistics, may be drawn from the results shown in Figure 2 and the stability analysis presented in paper (Kuravsky & Malykh, 2003):

- during preschool development, the systematic promotion of GIQ decrease is, as a rule, greater than the promotion of GIQ increase;
- during school development, the systematic promotion of GIQ decrease is, on the contrary, much less than the promotion of GIQ increase;
- the effect of systematic promotion of GIQ increase during children's school development is, as a rule, much greater than this effect during preschool development;
- during school development, the GIQ decrease flow rate is negligible for the IQ values that are less than the average GIQ;

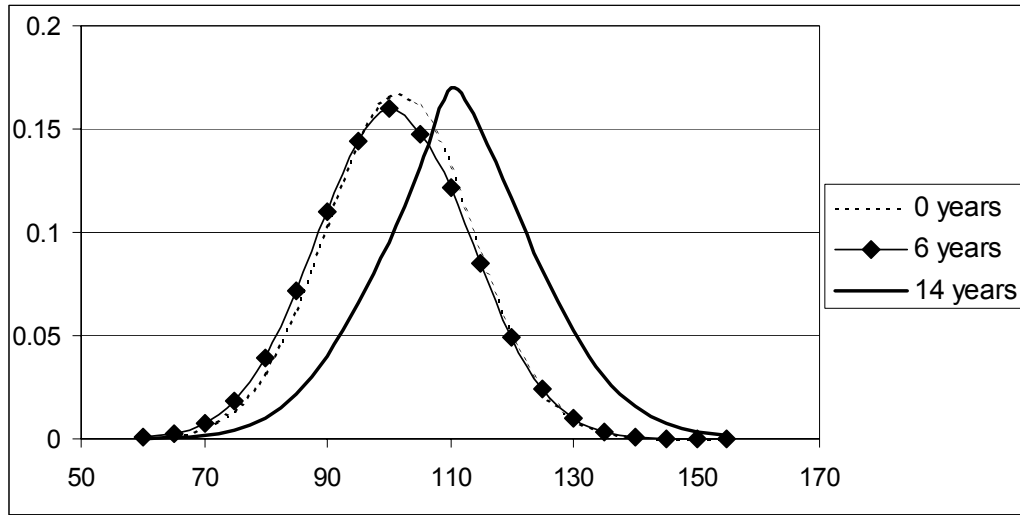


Figure 4. Envelopes of distributions of expected state probabilities at the moment of birth and test time points.

- e) during school development, the GIQ decrease flow rate is significant for the IQ values that are greater than the average GIQ;
- f) during school development, the GIQ increase flow rate for above the average IQ values is less than one for below the average IQs;
- g) during school development, the GIQ decrease flow rate for above the average IQ values is much greater than one for below the average IQs;
- h) during preschool development, the increase and decrease scores do not depend significantly on the IQ level.

These conclusions confirm general expectations to the effect that standard education does not promote development of capable children. However, the aforementioned *t*-test (see “Results”) shows the efficiency of Russian school education on the average: mean values after entering school became significantly greater. At the same time, preschool development does not yield essential shift in means: even if the expected value of m_0 would result from direct point estimate by some sample of 10 times greater size than the sample size for age 6, there would be no statistically significant differences between two means according *t*-test.

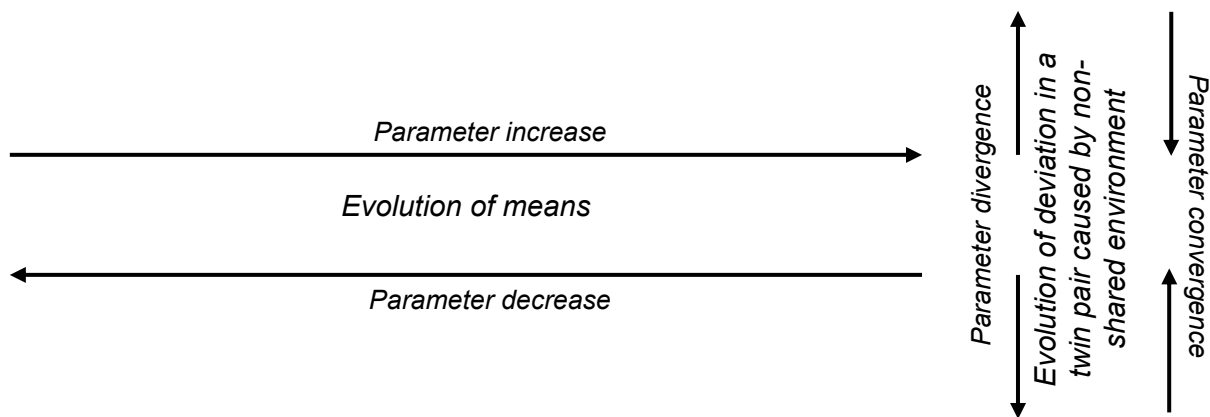


Figure 5. Two-dimensional representation of dynamics of individual characteristics.

It is of interest to compare obtained levels of increase and decrease flow rates discussed here and levels of convergence and divergence flow rates, which are rode on twin non-shared environment and studied in the paper (Gutman, 1954), for the same 5-unit IQ state intervals. In fact, it means to consider a 2-dimensional model describing dynamics of two independent characteristics (Figure 5). Juxtaposition with data of Table 3 shows that only the increased flow rates during school development may be significantly greater than the rates induced by twin non-shared environment. Sometimes, decreased and increased flow rates during preschool development are comparable with non-shared environment rates. At the same time, decrease characteristics during school development are much less. These facts demonstrate the important role of random undirected influences on population.

To appreciate the research effectiveness of the presented method in comparison with the allied approaches, the sample under study was examined with the aid of classical phenotypic simplex model, which is traditionally used in analysis of repeated measures. Following the idea of this model, variables η_1 and η_2 represent observed *IQ* measurements at ages 6 and 14. Their relationship is expressed by the first-order autoregression equation:

$$\eta_2 = \beta_2 \eta_1 + \zeta_2,$$

where β_2 is the regression of the observed variable at age 14 on the previous variable, ζ_2 represents a random input term (innovation⁵) that is uncorrelated with η_1 . This simplex model is illustrated graphically in the path diagram (Neal & Cardon, 1992; Jöreskog, 1970) in Figure 6.

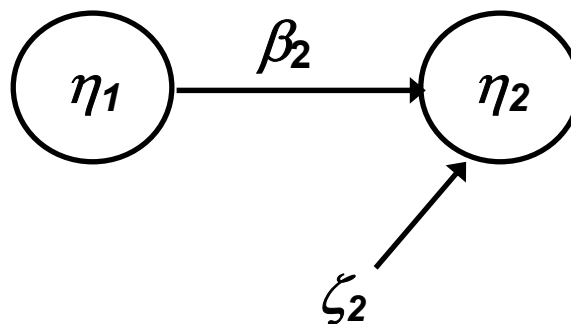


Figure 6. Two-point simplex model for studying *IQ* evolution: variables η_1 and η_2 represent observed *IQ* measurements at ages 6 and 14; β_2 is the regression of the observed variable at age 14 on the previous variable; ζ_2 represents an innovation at age 14.

⁵ The innovation is that part of the variable at the current time point that is not caused by the variable at the previous discrete time point. It differs from random measurement errors that do not influence subsequent observed variables. Measurement errors, which are frequently included in the classical simplex models, are not considered here.

The model in question has three free parameters: η_1 , ζ_2 , and β_2 . For two time points of observed measurements, the expected covariance matrix of variables η_1 and η_2 equals:

$$\begin{pmatrix} \text{var } \eta_1 & \beta_2 \text{ var } \eta_1 \\ \beta_2 \text{ var } \eta_1 & \beta_2^2 \text{ var } \eta_1 + \text{var } \zeta_2 \end{pmatrix}.$$

Since the number of free parameters is equal to the number of independent statistics in the observed covariance matrix, fitting the expected matrix to the observed one yields the perfect accordance and the following estimations: $\text{var } \eta_1=117$, $\text{var } \zeta_2=122$, $\text{var } \beta_2=0.7$.

Both the innovation and regression coefficient are statistically significant: provided that each of them taken separately equals to zero, goodness-of-fit measure in the form of maximum likelihood function, which is distributed as a chi-square under some general conditions (Bollen, 1989), gets statistically significant increase for one degree of freedom.

Sufficiently great proportion (68%) of observed variance at age 14 is due to innovation. The original variance at the first time point explains approximately two times less part - 32%. This is all scanty information, which may be obtained from the discussed covariance structure. It is much less than we can obtain with the aid of the proposed technique.

Of course, more observation points might be taken and subtler analysis might be carried out, but it is well known that acquisition of experimental data is more labor-consuming and expensive process than data analysis. Markov model helps us not only to learn more about reasons for phenotype changes, but extrapolate observed data to inaccessible time points (all points between zero age and age 14). However, using simplex and Markov models, we analyze different characteristics: flow rates versus proportions of observed variances explained by different factors. Markov models in their current form do not give the opportunity to analyze covariance (correlation) structures.

So, on the same observed data, Markov models, as a rule, yield more detailed analysis than simplex models.

One additional point should be also discussed here. We have considered the model that is truly longitudinal (i.e., measurements at observation moments are correlated) but only examined and modeled data that are cross-sectional (i.e., different and uncorrelated samples are used for different moments). We solved the problem by considering only frequencies within state intervals at each time. However, the more general case might be studied for sufficiently large samples, viz.: when the data of interest were a matrix of transition frequencies (cell i,j would contain the observed number of children changing from IQ state interval i at observed moment 1 to IQ state interval j at moment 2). The developed model would apply very nicely to this kind of data.

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