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**Araştırma Makalesi**

# Power of Some Statistical Tests for the Detection of Major Genes in Quantitative Traits: II. Tests of Normality

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**Abstract**

The power of some statistical methods to detect major genes controlling quantitative traits was evaluated using simulated data. The data were simulated according to a balanced half-sib family structure. One hundred and twenty eight scenario of major gene segregation based on all possible combinations from 4 levels of polygenic heritability (0.2, 0.4, 0.6 and 0.8), 2 modes of inheritance (additive and dominant), 4 levels of gene frequency (0.2, 0.4, 0.6 and 0.8) and 4 levels of major gene effect in phenotypic standard deviation ( $\sigma_P$ =0.5, 1, 2 and 3) were considered. The powers of 7 normality tests were compared for the detection of major genes. As tests of normality, power of tests of skewness and kurtosis coefficients, Bowman-Shenton, Shapiro-Wilk, Kolmogorov-Smirnov, Cramer-von Mises and Anderson-Darling normality tests were evaluated. The results showed that the power of all tests advanced with the increase of gene effects and the determination of dominant genes was easier than additive ones. The best power was obtained from Shapiro-Wilk, Bowman-Shenton and Anderson-Darling normality tests. In conclusion, these simple tests could be used in a systematic way as first indicators of major gene segregation in animal populations.

**Key words:** Quantitative traits, major gene, detection, normality test, simulation

## Kantitatif Karakterlere Etkili Major Genlerin Belirlenmesi Bakımından Kimi İstatistiki Testlerin Güçleri: II. Normal Dağılışa Uyum Testleri

**Özet**

Kantitatif karakterlere etkili major genlerin belirlenmesi bakımından kimi istatistiki metotların güçleri simülasyonla türetilen veriler kullanılarak değerlendirilmiştir. Veriler, dengeli bir üvey-kardeş familya yapısına göre türetilmiştir. Poligenik kalıtım derecesinin 4 düzeyi (0,2, 0,4, 0,6 ve 0,8), iki farklı kalıtım tarzı (kodominant ve dominant), 4 farklı gen frekansı düzeyi (0,2, 0,4, 0,6 ve 0,8) ve 4 farklı gen etkisi düzeyinin ( $\sigma_P$ = 0,5, 1,0, 2,0 ve 3,0) tüm olası kombinasyonuna dayanan toplam 128 farklı major gen açılımı senaryosu değerlendirilmiştir. Major genlerin belirlenmesi amacıyla farklı 7 normal dağılışa uyum (normalite) ve 4 familya-içi varyans homojenliği testinin güçleri karşılaştırılmıştır. Normalite testi olarak, çarpıklık ve diklik katsayılarının testi ile Bowman-Shenton, Shapiro-Wilk, Kolmogorov-Smirnov, Cramer-von Mises ve Anderson-Darling testlerinin güçleri belirlenmiştir. Sonuçlar gen etkisindeki yükselme ile birlikte tüm testlerin gücünün arttığını ve dominant etkili genlerin belirlenmesinin kodominant etkili genlerden çok daha kolay olduğunu ortaya koymuştur. Değerlendirilen testler içerisinde en yüksek güç sırasıyla Shapiro-Wilk, Bowman-Shenton ve Anderson-Darling normalite testlerinden elde edilmiştir. Sonuç olarak, bu basit metotlar hayvan populasyonlarında major genlerin açılımının ilk göstergesi olarak kullanılabilirler.

**Anahtar kelimeler:** Kantitatif karakterler, major gen, belirleme, normalite testi, simülasyon

## Introduction

According to quantitative theory, most traits of economic importance in livestock are assumed to be controlled by many genes each having a small effect. This theory has been successfully applied in animal breeding up to date. However, in the last two decade, several major genes or quantitative trait loci (QTL) having an important effect on quantitative traits has been identified in farm animals. Major genes or QTL with large effects are loci that have a large effect on the phenotypic appearance of the trait (Roberts and Smith, 1982; Miyake et al., 1999). The Booroola and Inverdale genes affecting ovulation rate (Piper and Bindon, 1982; Davis et al., 1988) and the callipyge gene affecting meat production in sheep (Cockett et al., 1993), the double muscling gene affecting meat production in cattle (Hanset and Michaux, 1985a,b), the halothane sensitivity and the RN gene affecting meat quality (Le Roy et al., 1990), the estrogen receptor locus affecting litter size in pigs (Rothschild et al., 1996), and the naked neck gene affecting heat tolerance and dwarf gene affecting body size in poultry (Merat, 1990) are the notable examples. The efficiency of classical selection programs can be improved by the use of major gene information. For this reason, it is of great interests to detect such genes and genotyping individuals. With the advances

of molecular genetics and statistical methods in the last years, detection of major genes or QTL has been more possible. However, detection of major genes without molecular genetic applications remains important due to difficulties and high costs of molecular application. Furthermore, in livestock populations phenotypic observations are often abundantly available at a low cost and it is worthwhile to use them to realize a statistical analysis for searching major gene effects.

If a major gene, whose effect is large enough, segregates there will be a detectable departure from normality (Le Roy and Elsen, 1992; Cemal, 1996; Falconer and Mackay, 1996). Therefore, tests of normality can be used for determination of major genes as a first indicator. Simple indicators of major genes have been suggested elsewhere (Le Roy and Elsen, 1992). But, more detailed properties of normality tests for the detection of major genes were not studied until now. From this point of view the present paper aimed to evaluate power of a number of normality tests for the detection of major genes.

## Materials and Methods

### *Experimental Design*

To determine the power of statistical tests for the identification of major genes, polygenic and mixed (polygenes + a major gene) inheritance models were compared. The polygenic data were simulated according to a balanced half-sib family structure: one data set consists of 50 sire families with 20 dams per sire and one progeny per dam. One phenotypic observation was generated for each progeny. Sires and dams are assumed to be unrelated. The model to describe the data based on polygenic inheritance can be represented as:

$$y_{ij} = \mu + s_i + e_{ij}$$

where $y_{ij}$ is the observation of j$^{th}$ progeny of i$^{th}$ sire, $\mu$ is the overall population mean of the polygenic and environmental components (set to zero), $s_i$ is the random effect of i$^{th}$ sire (i.e. polygenic component) and $e_{ij}$ is the residual random effect for each progeny.

For progenies the true breeding values were obtained from a normal distribution with mean zero and variance $\sigma_a^2 = (¼\, h^2)\sigma_P^2$ where phenotypic variance ($\sigma_P^2$) was set equal to 1. Their residual values were generated from a normal distribution with mean zero and variance $\sigma_e^2 = (1 - ¼\, h^2)\sigma_P^2$. Afterwards, the phenotypic value of each progeny was obtained as the sum of the true breeding value (~N(0, $\sigma_a^2$)) and the residual value (~N(0, $\sigma_e^2$)) where N represents the normal distribution. By this way, 4 separate data sets each contain 100 replicates were simulated for different values of polygenic heritability ($h^2$= 0.2, 0.4, 0.6 and 0.8).

A single major gene with two alleles (*A* and *a*) was considered. There are three genotypes, *AA*, *Aa*, and *aa*, taking genetic value as *a*, *d*, and –*a*, respectively, where *a* is the additive and *d* is the dominant genetic effect. The effect of a major gene in phenotypic standard deviation ($\sigma_P$) unit was considered as the difference between the means of two homozygote genotypes ($2a = \mu_{AA} - \mu_{aa}$). The dominance of the major gene was interpreted by $d = \mu_{Aa} - (\mu_{AA} + \mu_{aa})/2$. The parameter set up used for all tests was as the following: polygenic heritability ($h^2 = 4\sigma_a^2 / \sigma_P^2$) took values of 0.2, 0.4, 0.6, or 0.8; type of dominance took values of *d*=0 (additive or codominant), or *d*=*a* (complete dominance); frequency of the major gene p*(A)* took values of 0.2, 0.4, 0.6, or 0.8; and magnitude of major gene effect as difference of means of two homozygotes in $\sigma_P$ unit took values of 0.5, 1, 2, or 3. In this way 128 different cases of major gene segregation based on all possible combinations from 4 levels of polygenic heritability, 2 modes of inheritance, 4 levels of gene frequency and 4 levels of major gene effect were evaluated with various tests of normality.

For parents, the genotypes of the major gene were calculated from given allele frequency. Then the genotype of progenies assigned from their parent's genotypes. Major gene effects were added to polygenic data of progenies according to their genotypes using uniform random numbers. Consequently, polygenic effects and major gene effect was combined in the following statistical model to obtain mixed (polygenes + a major gene) data:

$$y_{ij}^k = \mu_k + s_i + e_{ij}$$

where $y_{ij}^k$ is the observation of j$^{th}$ progeny of i$^{th}$ sire with major genotype *k* (*AA, Aa* and *aa*), $\mu_k$ is the mean value of the performances of genotype *k* progeny, $s_i$ is the random effect of i$^{th}$ sire (i.e. polygenic component) and $e_{ij}$ is the residual random effect.

It is assumed that $H_0$ and $H_1$ are the hypotheses of polygenic and mixed (polygenes + a major gene) inheritance, respectively. Under $H_0$ we consider a trait is normally distributed as a result of polygenic inheritance.

For each of the test statistics power represents probability of rejecting the null hypothesis when the alternative hypothesis is true. The power of tests at the

5% error level was estimated for each situation studied by taking the number of test statistic values that exceeded the corresponding $H_0$ quantile. The power of all tests was estimated from 100 replications. The robustness of the test statistics was not examined here.

A macro was written in SAS Macro Language for the simulation of data sets and statistical analyses and all simulations and analyses were performed by SAS software (SAS, 1999a,b).

### *Statistical tests and analyses*

When a major gene segregates the distribution of phenotypes for each of the major-locus genotypes will be normal. But the resulting phenotypic distribution, a mixture of normals, can exhibit non-normality (Falconer and Mackay, 1996; Lynch and Walsh, 1997). A variety of tests for normality have been developed. In this study, seven of which were evaluated for the detection of major gene segregation. These tests are skewness and kurtosis coefficients, Bowman-Shenton, Shapiro-Wilk, Kolmogorov-Smirnov, Anderson-Darling and Cramer-von Mises normality tests (Shapiro and Wilk, 1965; Bowman and Shenton, 1975; Snedecor and Cochran, 1989; Lynch and Walsh, 1997). Details of evaluated tests are given as follows:

*Test of skewness coefficient (Skw).* Observations that are normally distributed should have a skewness near zero. Its deviation from zero is due to departure from normality. The sample estimate of skewness parameter ( $\gamma_3$ ) is computed as (Yıldız et al., 1998):

$$\gamma_3 = \frac{\sum (y_{ij} - \mu)^3 / n}{\sigma^3}$$

where $y_{ij}$ is the observation of j[th] progeny of i[th] sire, $\mu$ is the overall mean, n is the sample size and $\sigma$. is the standard deviation of the sample.

The difference ( $\Delta\gamma_3$ ) between the estimates of skewness for normally distributed polygenic data and the data including a major gene effect was statistically tested for the detection of a major gene effect. For large sample sizes (n>150), skewness coefficient is approximately normally distributed with mean 0 and standard deviation $sd(\gamma_3) = \sqrt{6/n}$ (Düzgüneş et al., 1983; Snedecor and Cochran, 1989). In this case, estimated test statistics $z_e = \Delta\gamma_3 / sd(\gamma_3)$ were compared with two tailed $z_{\alpha/2}$ score for the rejection of the null hypothesis ($H_0$: $\Delta\gamma_3 = 0$) that explain polygenic inheritance.

*Test of kurtosis coefficient (Kur).* Kurtosis is another criterion of a distribution's departure from normality. Observations that are normally distributed should have a kurtosis near zero and significance deviation from this value indicates the non-normality of data. The sample estimate of this coefficient is denoted by $\gamma_4$ and computed as (Yıldız et al., 1998):

$$\gamma_4 = \frac{\sum (y_{ij} - \mu)^4 / n}{\sigma^4} - 3$$

where $y_{ij}$ is the observation of j[th] progeny of i[th] sire, $\mu$ is the overall mean, n is the sample size and $\sigma$. is the standard deviation of the sample.

The difference ( $\Delta\gamma_4$ ) between the estimates of kurtosis for normally distributed polygenic data and the data including a major gene effect were statistically tested for the detection of a major gene effect. If the sample size is greater than 1000, kurtosis coefficient is approximately normally distributed with mean 0 and standard deviation, $sd(\gamma_4) = \sqrt{24/n}$ (Düzgüneş et al., 1983; Snedecor and Cochran, 1989).

In this case, estimated test statistics $z_e = \Delta\gamma_4 / sd(\gamma_4)$ were compared with two tailed $z_{\alpha/2}$ score for the rejection of the null hypothesis ($H_0$: $\Delta\gamma_3 = 0$) that explain polygenic inheritance.

*Bowman-Shenton test (B-S).* Deviation from normality indicates significant skewness and/or kurtosis, both of which have expected value zero under the assumption of normality. Bowman and Shenton (Bowman and Shenton, 1975) proposed a joint test of normality. This statistic is defined as:

$$BS = \frac{n \cdot \gamma_3^2}{6} + \frac{n \cdot \gamma_4^2}{24}$$

where n is the sample size, $\gamma_3$ and $\gamma_4$ are the standardized sample skewness and kurtosis, respectively. For larger sample sizes, *BS* is distributed as a $\chi^2$ with two degrees of freedom. Thus, the hypothesis that a distribution is normal is rejected at the 5% level if *BS*>5.99.

*Shapiro-Wilk test (S-W).* The Shapiro-Wilk W statistic is computed only when the number of observations (n) is less then or equal to 2000. The W statistic is the ratio of the best estimator of variance to the usual corrected sum of squares estimator of variance (Shapiro and Wilk, 1965). The W statistic for normality defined by:

$$W = \left(\sum_{i=1}^{n} a_i y_i\right)^2 \Big/ \sum_{i=1}^{n} (y_i - \bar{y})^2$$

where n is the number of observations, $a_i$ is the tabulated coefficients, $y_i$ is the observation of i[th] individual and $\bar{y}$ is the mean of the observations.

Small values of W lead to the rejection of the null ($H_0$) hypothesis of normality. The method for computing the *p*-value (the probability of obtaining a W statistic less than or equal to observed value) depends on n. For $12 \le n \le 2000$, simulation results are used to get the approximate normalizing transformation (Royston, 1992):

$$Z_n = (\log(1 - W) - \mu)/\sigma$$

where $Z_n$ is a standard normal variate and the values of $\sigma$ and $\mu$ are functions of n obtained from simulation results.

Large values of $Z_n$ indicate departure from normality (Shapiro and Wilk, 1965).

*EDF tests for normality*. The Kolmogorov-Smirnov, Anderson-Darling and Cramer-von Mises tests for normality are based on the empirical distribution function, EDF (Stephens, 1974). These tests are often referred to as EDF tests and are based on various measures of the discrepancy between the empirical distribution function $F_n(x)$ and the hypothesized cumulative distribution function $F(x)$. Under the null hypothesis, $F(x)$ is the normal distribution. The empirical distribution function is defined as:

$$F_n(x) = \begin{cases} 0, & x < X_{(1)} \\ i/n, & X_{(i)} \le x < X_{(i+1)} \quad i = 1,...,n-1 \\ 1, & X_{(n)} \le x \end{cases}$$

The computational formulas for the EDF statistics use the probability integral transformation $U = F(x)$. If $F(x)$ is the distribution function of $X$, the random variable $U$ is uniformly distributed between 0 and 1. Given n observations $X_{(1)},...,X_{(n)}$, the values $U_{(i)} = F(x_{(i)})$ are computed. These values are used to compute EDF test statistics as follows (Stephens, 1974):

*(a) Kolmogorov-Smirnov test (K-S).*The Kolmogorov-Smirnov statistic D belongs to the supremum class of EDF statistics. The statistic is defined as:

$$D = \sup_x |F_n(x) - F(x)|$$

This statistic is computed as the maximum of $D^+$ and $D^-$.

$$D^+ = \max_i \left(\frac{i}{n} - U_{(i)}\right)$$

$$D^- = \max_i \left(U_{(i)} - \frac{i-1}{n}\right)$$

$$D = \max(D^+, D^-)$$

*(b) Anderson-Darling test (A-D).* The Anderson-Darling $A^2$ statistic and the Cramer-von Mises statistic belong to the quadratic class of EDF statistics. This class of statistics is based on the squared difference $(F_n(x)-F(x))^2$. The statistic is computed as:

$$A^2 = -n - \frac{1}{n}\sum_{i=1}^{n}\left[(2i-1)\left(\log U_{(i)} + \log\left(1 - U_{(n+1-i)}\right)\right)\right]$$

*(c) Cramer-von Mises test (CvM).* The Cramer-von Mises statistic is computed as:

$$W^2 = \sum_{i=1}^{n}\left(U_{(i)} - \frac{2i-1}{2n}\right)^2 + \frac{1}{12n}$$

## Results

Simulation results on the power of normality tests for detection of major genes were given under separate headings according to mode of major gene inheritance (additive and dominant).

### *Power of the tests for detection of additive major genes*

The power (%) of normality tests for the detection of additive major genes with 0.5, 1.0, 2.0 and 3.0 $\sigma_P$ of gene effect are given in Table 1. These results showed that the power of all tests increased with the increase of gene effect from 0.5 to 3 $\sigma_P$. But the level of polygenic heritability does not have an obvious effect on the power of tests.

The performances of all tests were quite low (≤4%) for the detection of additive major genes with 0.5 $\sigma_P$ of gene effect. Among the evaluated tests, the power of tests of skewness and kurtosis coefficients were about 0%. The power of the tests was not affected by the different frequencies of major genes.

The power of tests increased (≤11%) with the increase of gene effect to 1 $\sigma_P$ and were not affected by different frequencies of major genes. The Kolmogorov-

Table 1. The power (%) of various normality tests for the detection of additive major genes with different magnitude of gene effects (0.5 to 3.0 $\sigma_P$)

| Polygenic $h^2$ | Major gene frequency | Magnitude of Major Gene Effect | | | | | | | | | | | | | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | 0.5 $\sigma_P$ | | | | | | | 1.0 $\sigma_P$ | | | | | | | 2.0 $\sigma_P$ | | | | | | | 3.0 $\sigma_P$ | | | | | | |
| | | Skw | Kur | B-S | S-W | K-S | CvM | A-D | Skw | Kur | B-S | S-W | K-S | CvM | A-D | Skw | Kur | B-S | S-W | K-S | CvM | A-D | Skw | Kur | B-S | S-W | K-S | CvM | A-D |
| 0.20 | 0.2 | 0 | 0 | 1 | 2 | 0 | 1 | 2 | 0 | 0 | 2 | 1 | 4 | 5 | 4 | 37 | 5 | 28 | 22 | 14 | 16 | 17 | 99 | 12 | 93 | 97 | 83 | 91 | 98 |
| | 0.4 | 0 | 0 | 2 | 0 | 1 | 0 | 0 | 0 | 0 | 2 | 2 | 11 | 5 | 4 | 8 | 7 | 10 | 13 | 9 | 12 | 9 | 25 | 27 | 40 | 44 | 22 | 29 | 33 |
| | 0.6 | 0 | 0 | 0 | 0 | 0 | 0 | 2 | 0 | 0 | 2 | 3 | 3 | 3 | 5 | 6 | 5 | 4 | 3 | 7 | 6 | 5 | 33 | 29 | 37 | 36 | 30 | 35 | 39 |
| | 0.8 | 0 | 0 | 4 | 2 | 2 | 2 | 2 | 0 | 0 | 1 | 1 | 1 | 2 | 2 | 39 | 3 | 25 | 30 | 22 | 21 | 27 | 98 | 10 | 93 | 96 | 82 | 92 | 95 |
| 0.40 | 0.2 | 0 | 0 | 0 | 0 | 1 | 2 | 2 | 0 | 0 | 3 | 3 | 4 | 5 | 6 | 33 | 4 | 24 | 30 | 19 | 25 | 25 | 97 | 10 | 97 | 98 | 84 | 90 | 94 |
| | 0.4 | 0 | 0 | 2 | 2 | 4 | 1 | 2 | 0 | 0 | 1 | 4 | 6 | 3 | 3 | 7 | 8 | 4 | 7 | 10 | 9 | 10 | 26 | 29 | 41 | 47 | 21 | 29 | 39 |
| | 0.6 | 0 | 0 | 1 | 2 | 4 | 3 | 1 | 0 | 0 | 2 | 1 | 3 | 1 | 1 | 13 | 6 | 8 | 12 | 9 | 10 | 10 | 32 | 33 | 43 | 46 | 31 | 31 | 33 |
| | 0.8 | 0 | 0 | 2 | 1 | 2 | 1 | 2 | 0 | 0 | 1 | 2 | 2 | 3 | 4 | 31 | 4 | 25 | 29 | 21 | 17 | 21 | 99 | 13 | 95 | 96 | 81 | 90 | 94 |
| 0.60 | 0.2 | 0 | 0 | 1 | 1 | 1 | 1 | 1 | 0 | 0 | 3 | 2 | 7 | 2 | 2 | 36 | 5 | 25 | 29 | 25 | 23 | 30 | 95 | 9 | 96 | 96 | 77 | 90 | 94 |
| | 0.4 | 0 | 0 | 1 | 2 | 2 | 2 | 1 | 0 | 0 | 2 | 3 | 4 | 5 | 3 | 4 | 6 | 11 | 10 | 17 | 13 | 11 | 23 | 27 | 48 | 50 | 27 | 41 | 43 |
| | 0.6 | 0 | 0 | 3 | 0 | 0 | 2 | 1 | 0 | 0 | 2 | 2 | 2 | 2 | 2 | 5 | 4 | 5 | 9 | 11 | 6 | 7 | 22 | 30 | 47 | 54 | 26 | 34 | 39 |
| | 0.8 | 0 | 0 | 0 | 0 | 0 | 1 | 1 | 0 | 0 | 2 | 1 | 2 | 1 | 1 | 32 | 6 | 26 | 25 | 21 | 25 | 24 | 95 | 8 | 96 | 96 | 75 | 93 | 95 |
| 0.80 | 0.2 | 0 | 0 | 0 | 0 | 4 | 0 | 0 | 0 | 0 | 1 | 0 | 1 | 3 | 3 | 38 | 2 | 26 | 33 | 16 | 27 | 30 | 99 | 10 | 94 | 95 | 71 | 87 | 88 |
| | 0.4 | 0 | 0 | 0 | 1 | 2 | 2 | 1 | 1 | 1 | 2 | 2 | 5 | 6 | 5 | 11 | 6 | 9 | 11 | 8 | 9 | 10 | 31 | 24 | 42 | 45 | 31 | 34 | 41 |
| | 0.6 | 0 | 0 | 0 | 1 | 2 | 0 | 0 | 0 | 0 | 2 | 2 | 4 | 4 | 3 | 5 | 3 | 4 | 8 | 7 | 10 | 10 | 36 | 28 | 52 | 59 | 21 | 33 | 40 |
| | 0.8 | 0 | 0 | 1 | 1 | 2 | 3 | 4 | 1 | 0 | 0 | 3 | 3 | 4 | 3 | 35 | 2 | 20 | 21 | 18 | 17 | 22 | 97 | 12 | 94 | 93 | 76 | 86 | 90 |

**Abbreviations:** $h^2$: heritability, $\sigma_P$ : phenotypic standard deviation, Skw: Skewness, Kur: Kurtosis, B-S: Bowman-Shenton, S-W: Shapiro-Wilk, K-S: Kolmogorov-Smirnov, CvM: Cramer-von Mises, A-D: Anderson-Darling

Smirnov, Cramer-von Mises and Anderson-Darling tests are more effective than other normality tests at this level of gene effect.

All tests' power was significantly increased (≤39%) when effect of major gene increased to $2\sigma_P$. The power of test of skewness coefficients was dramatically increased. All normality tests except test of kurtosis coefficients were more powerful for extreme gene frequencies (p=0.2 or 0.8) than moderate (p=0.4 or 0.6).

When the additive major gene have a gene effect of $3\sigma_P$, power of all normality tests except test of kurtosis coefficients were exceeded 50%. The power of all normality tests except test of kurtosis coefficient were higher for extreme gene frequencies (p=0.2 or 0.8) than intermediate (p=0.4 or 0.6) as in the case of segregation of major genes with an effect of 2 $\sigma_P$. In contrary, the test of kurtosis coefficients was powerful in the segregation of major genes with intermediate frequencies. The power of test of skewness coefficients, Bowman-Shenton, Shapiro-Wilk, Cramer-von Mises and Anderson-Darling normality tests are fairly high (over the 90%) in extreme gene frequencies.

### *Power of the tests for detection of completely dominant major genes*

The power (%) of normality tests for the detection of dominant genes with 0.5, 1, 2 and $3\sigma_P$ of major gene effects are presented in Table 2. As in the case of additive major gene segregation, power of the all studied tests for detection of dominant major genes were not significantly affected by level of polygenic heritability. The power of all tests increased with the increase of gene effect from 0.5 to $3\sigma_P$. The performances of all tests were very low for the detection of major genes with 0.5 and 1 $\sigma_P$ of gene effect. The power of tests, especially for dominant genes, was increased suddenly for a gene effect of 2 or $3\sigma_P$.

The power of all tests was fairly low for the detection of major genes with $0.5\sigma_P$ of gene effect. Only the Kolmogorov-Smirnov test was reached to a power of 7% in one situation. But, the power of all of other tests was smaller than or equal to 4%. The power of the tests were not changed with the increase of frequency of major genes. Among the evaluated tests, the power of tests of skewness and kurtosis coefficients were exactly 0%.

With the augmentation of major gene effect from 0.5 to $1\sigma_P$ the power of all tests partially increased (≤12%). Tests were more powerful in low or moderate frequencies. The Bowman-Shenton, Shapiro-Wilk, Kolmogorov-Smirnov, Cramer-von Mises and Anderson-Darling are more powerful normality tests for a major gene effect of $1\sigma_P$.

A sharp increase in the power of tests was observed when the magnitude of segregating dominant major gene effect increased from 1 to $2\sigma_P$. In most of the situations, in particularly at major gene frequencies of 0.2 and 0.6, power of tests was about 100%. Tests appear less powerful when the frequency of major gene is 0.8.

In most cases of the existence of dominant major genes with an effect of $3\sigma_P$, the power of the tests reached to approximately 100%. The influences of major gene frequency on power of the normality tests were disappeared with the increase of magnitude of major gene effect to $3\sigma_P$.

### *Mean power of the tests*

To clarify the results, power of the tests at the 4 level of gene frequency and 4 level of polygenic heritability were joined in one mean to obtain average power of the each test. Mean power of the tests for the detection of additive and dominant major genes with different level of gene effects (0.5 to 3 σ) are given in Figure 1.

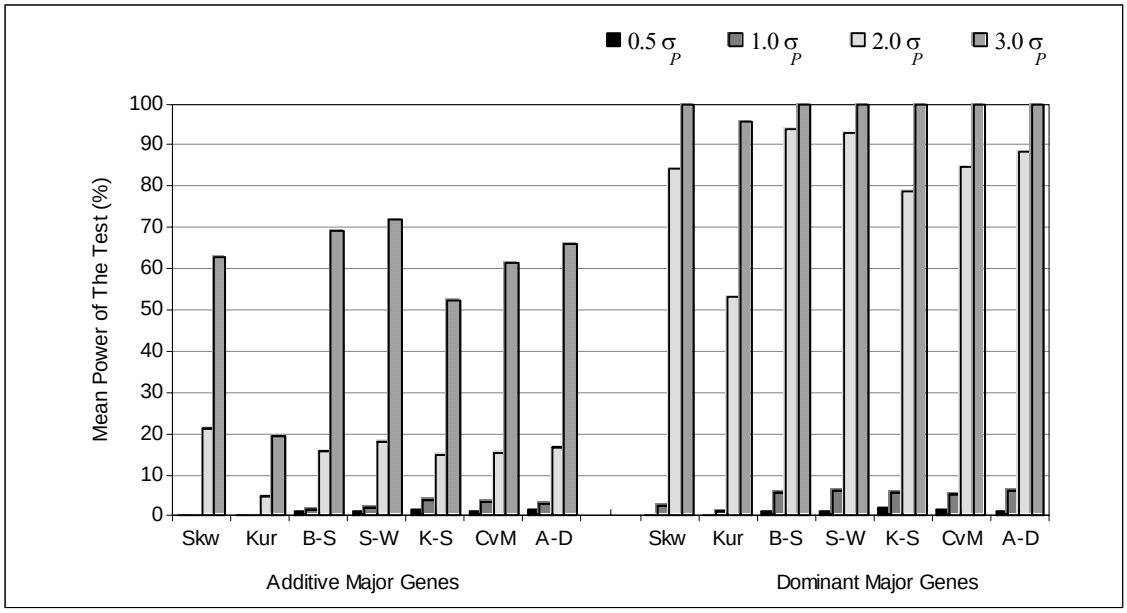
In the case of segregation of additive genes, all of the tests except kurtosis test gave a mean power above 50% for a gene effect of $3\sigma_P$. With a gene effect of $\leq 2\sigma_P$, none of tests had an average power greater than about 22%. Among the normality tests studied, the best power for the detection of additive genes was obtained from Shapiro-Wilk, Bowman-Shenton and Anderson-Darling tests, respectively.

In the existence of dominant genes, all test statistics based on normality had a mean power greater than 50% for a gene effect of $2\sigma_P$ and power reached approximately 100% for a gene effect of $3\sigma_P$. The Bowman-Shenton, Shapiro-Wilk and Anderson-Darling are the three more powerful tests among normality tests in the existence of dominant genes. Power of tests were rather high for detection of dominant genes than additive ones when the magnitude of major gene are high (2 or 3 $\sigma_P$).

Table 2. The power (%) of various normality tests for the detection of dominant major genes with different magnitude of gene effects (0.5 to 3.0 σP)

| Polygenic $h^2$ | Major gene frequency | Magnitude of Major Gene Effect | | | | | | | | | | | | | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | 0.5 $\sigma_P$ | | | | | | | 1.0 $\sigma_P$ | | | | | | | 2.0 $\sigma_P$ | | | | | | | 3.0 $\sigma_P$ | | | | | | |
| | | Skw | Kur | B-S | S-W | K-S | CvM | A-D | Skw | Kur | B-S | S-W | K-S | CvM | A-D | Skw | Kur | B-S | S-W | K-S | CvM | A-D | Skw | Kur | B-S | S-W | K-S | CvM | A-D |
| 0.20 | 0.2 | 0 | 0 | 2 | 1 | 2 | 3 | 3 | 0 | 3 | 7 | 7 | 7 | 10 | 9 | 78 | 65 | 97 | 98 | 97 | 98 | 99 | 99 | 100 | 100 | 100 | 100 | 100 | 100 |
| | 0.4 | 0 | 0 | 0 | 0 | 4 | 1 | 0 | 3 | 1 | 4 | 3 | 8 | 6 | 7 | 75 | 65 | 94 | 98 | 95 | 99 | 99 | 100 | 100 | 100 | 100 | 100 | 100 | 100 |
| | 0.6 | 0 | 0 | 1 | 1 | 2 | 2 | 3 | 6 | 1 | 11 | 8 | 3 | 5 | 5 | 100 | 23 | 100 | 100 | 98 | 100 | 100 | 100 | 80 | 100 | 100 | 100 | 100 | 100 |
| | 0.8 | 0 | 0 | 3 | 0 | 0 | 0 | 0 | 0 | 0 | 3 | 4 | 2 | 3 | 2 | 87 | 56 | 81 | 74 | 28 | 35 | 53 | 100 | 100 | 100 | 100 | 100 | 100 | 100 |
| 0.40 | 0.2 | 0 | 0 | 1 | 3 | 4 | 4 | 2 | 0 | 0 | 4 | 10 | 6 | 8 | 10 | 71 | 72 | 100 | 100 | 99 | 100 | 100 | 99 | 100 | 100 | 100 | 100 | 100 | 100 |
| | 0.4 | 0 | 0 | 3 | 2 | 7 | 4 | 2 | 3 | 0 | 7 | 9 | 11 | 8 | 8 | 77 | 66 | 96 | 98 | 94 | 98 | 98 | 100 | 100 | 100 | 100 | 100 | 100 | 100 |
| | 0.6 | 0 | 0 | 2 | 2 | 0 | 1 | 1 | 5 | 0 | 8 | 8 | 6 | 5 | 8 | 100 | 18 | 99 | 99 | 96 | 98 | 100 | 100 | 84 | 100 | 100 | 100 | 100 | 100 |
| | 0.8 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 3 | 5 | 1 | 3 | 3 | 88 | 56 | 82 | 77 | 30 | 46 | 59 | 100 | 100 | 100 | 100 | 97 | 100 | 99 |
| 0.60 | 0.2 | 0 | 0 | 1 | 1 | 2 | 1 | 0 | 2 | 2 | 9 | 11 | 9 | 5 | 6 | 73 | 77 | 98 | 100 | 98 | 100 | 100 | 100 | 100 | 100 | 100 | 100 | 100 | 100 |
| | 0.4 | 0 | 0 | 2 | 2 | 2 | 2 | 2 | 1 | 4 | 3 | 3 | 4 | 5 | 4 | 79 | 63 | 99 | 99 | 93 | 98 | 98 | 100 | 100 | 100 | 100 | 100 | 100 | 100 |
| | 0.6 | 0 | 0 | 2 | 2 | 3 | 1 | 1 | 11 | 0 | 10 | 8 | 7 | 9 | 8 | 100 | 32 | 100 | 100 | 97 | 99 | 99 | 100 | 86 | 100 | 100 | 100 | 100 | 100 |
| | 0.8 | 0 | 0 | 0 | 1 | 1 | 0 | 0 | 0 | 0 | 4 | 1 | 0 | 0 | 0 | 87 | 62 | 80 | 73 | 38 | 50 | 54 | 100 | 100 | 100 | 100 | 99 | 100 | 100 |
| 0.80 | 0.2 | 0 | 0 | 0 | 1 | 4 | 1 | 1 | 6 | 2 | 5 | 7 | 10 | 12 | 12 | 74 | 59 | 98 | 99 | 81 | 96 | 98 | 100 | 100 | 100 | 100 | 100 | 100 | 100 |
| | 0.4 | 0 | 0 | 0 | 0 | 1 | 2 | 1 | 2 | 4 | 4 | 5 | 8 | 6 | 9 | 76 | 62 | 93 | 96 | 88 | 96 | 98 | 100 | 100 | 100 | 100 | 100 | 100 | 100 |
| | 0.6 | 0 | 0 | 1 | 0 | 2 | 1 | 1 | 4 | 1 | 5 | 6 | 6 | 1 | 3 | 100 | 18 | 100 | 100 | 99 | 100 | 100 | 100 | 80 | 100 | 100 | 100 | 100 | 100 |
| | 0.8 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 1 | 3 | 1 | 1 | 1 | 2 | 84 | 60 | 83 | 78 | 32 | 41 | 58 | 100 | 100 | 100 | 100 | 99 | 100 | 100 |

**Abbreviations:** $h^2$: heritability, $\sigma_P$ : phenotypic standard deviation, Skw: Skewness, Kur: Kurtosis, B-S: Bowman-Shenton, S-W: Shapiro-Wilk, K-S: Kolmogorov-Smirnov, CvM: Cramer-von Mises, A-D: Anderson-Darling




**Abbreviations:** $\sigma_P$: phenotypic standard deviation, Skw: Skewness, Kur: Kurtosis, B-S: Bowman-Shenton, S-W: Shapiro-Wilk, K-S: Kolmogorov-Smirnov, CvM: Cramer-von Mises, A-D: Anderson-Darling

Figure 1. Mean power (%) of the normality tests for the detection of additive and dominant major genes with different level of gene effects (0.5 to 3 $\sigma_P$).

## Discussion

The dominant major genes were determined more easily than additive ones in all scenarios of major gene segregation. Seemingly, dominant genes are determined easily than codominant ones in other studies using same or different tests (Le Roy, 1989; Knott and Haley, 1991; Janss and Van Der Werf, 1992; Le Roy and Elsen, 1992; Elsen and Le Roy, 1995).

When an additive major gene is segregating, all tests except kurtosis are more powerful for extreme gene frequencies (0.2 or 0.8) than moderate (0.4 or 0.6). But this phenomenon is not valid in the case of dominant genes.

Up to date, only the Shapiro-Wilk and Kolmogorov-Smirnov normality tests applied to actual data of RN gene in pigs (Le Roy, 1989), rate of milk flow gene in goats (Ricordeau et al., 1989) and double muscling gene in Belgian-Blue cattle (Hanset and Michaux, 1985a,b) and in all three situations segregation of major genes was confirmed by this tests.

The power of a different class of statistical tests including within-family variance homogeneity tests was investigated with same data sets by Cemal and Karaca (2005). The power of normality tests was quite higher than within-family variance homogeneity tests evaluated in mentioned study, especially for higher gene effects. In nearly all situations studied, the normality tests were found more powerful than within-family variance homogeneity tests.

An increased attention has been given to the detection of major loci or QTL in the last years. Simple statistical methods that compared in this paper could be used as a first indicator of major gene segregation in animal populations. Segregation of additive major genes with 3 or more and of dominant major genes with 2 or more $\sigma_P$ of gene effect may be easily determined by these simple tests. Checking data with more than one test of normality may be more meaningful due to different power of tests to various situations of major gene segregation. Concerning the power, the use of Bowman-Shenton, Shapiro-Wilk and Anderson-Darling normality tests are primarily recommended. However, if environmental variation is sufficiently high relative to the effects of any individual gene or if major alleles are at low frequency, the effects of segregating major genes

can be entirely obscured. Therefore, the effects of macro environmental components need to be removed from phenotypic data to make these tests more powerful. In addition to the normality tests, within-family variance homogeneity test may be used for checking phenotypic data of animals to detect major genes. When positive results obtained for any major genes based on these simple tests, these results would have to be confirmed and detailed by more complicated methods such as segregation analysis of phenotypic data or molecular genetic applications.

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