

Genetic and environmental determinants of dental occlusal variation in South Australian twins

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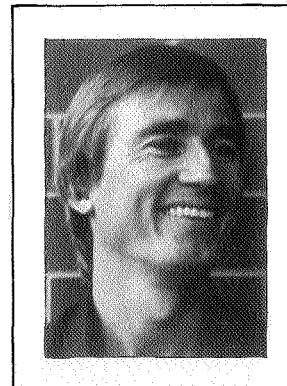
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Abstract : As part of an ongoing study of dentofacial morphology in South Australian twins, several occlusal traits were compared between forty eight monozygous and thirty four dizygous twin pairs, as well as estimating genetic variances for these features. Significant genetic variance was noted for incisal overbite and overjet, sagittal molar relationship, and also rotations and displacements of anterior teeth. However, heritability estimates were low in magnitude (about 30%) *emphasising the importance of environmental influences on occlusal variation.*

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Key words : Occlusal variation, twins, genetics.

INTRODUCTION

In contrast to most medical and dental problems that can be considered pathological, dental malocclusions usually result from variations in normal development for which there is no apparent cause (Proffit 1986). Indeed, the term 'occlusal variation' seems more appropriate than 'malocclusion', emphasising as it does the continuous range of occlusal relationships that we observe, with more severe cases being located at the extremes of the distribution (Smith and Bailit, 1977).

Although it is generally accepted that malocclusions of non-specific origin result from a combination of genetic and environmental influences during development, the relative importance of these factors remains unclear. Early traditional twin studies (Lundström 1948) and intra-familial comparisons (Stein et al. 1956) indicated that occlusal traits were under considerable genetic control, but more recent reports on twins (e.g. Potter, Corruccini and Green, 1981) and first-degree relatives (Harris and Smith, 1980) have emphasised the role of environmental factors. Anthropological studies, demonstrating rapid increases in occlusal variation in non-industrialised populations following the adoption of western-style food habits, also add support to the notion that non-genetic factors, possibly related to masticatory function, are important contributors to variation within the dentition (e.g. Corruccini 1984).

As Proffit (1986) has pointed out, history indicates that prevailing views about the causes of malocclusions have affected the type of treatment offered to patients to a surprising degree. This study provides the first estimates of genetic variance for occlusal traits in Australian twins, including tests of several basic assumptions underlying the twin model. A better understanding of the relative effects of genetic and environmental influences on different occlusal features should ultimately lead to improved preventive and treatment planning rationales in orthodontics.

MATERIALS AND METHODS

As part of an ongoing study of dentofacial structures in South Australian twins, several occlusal traits have been compared between a sample of 48 pairs of monozygous (MZ) twins and 34 dizygous (DZ) pairs, as well as estimating genetic variance of the study variables. The twins, comprising 84 males and 80 females, were aged between 13 and 26 years with an average age of 16.2 years. Only individuals with complete permanent dentitions on at least one side (apart from third molars) and no history of orthodontic treatment were included. Zygosity was determined by comparison of several genetic markers in the blood.

Occlusal traits were measured and scored directly from randomly-arranged dental models (co-twins scored separately) by one of us (RC), with opposing teeth in intercuspal position or centric occlusion. The measurement methods employed have been detailed previously (Corruccini and Potter, 1980; Sharma and Corruccini, 1986).

The following variables were defined. Overjet (OJ) was the horizontal distance from the labial surface of the best positioned $\bar{1}$ to the labial surface of $\bar{I}$ on the same side. Overbite (OB) was the proportion of $\bar{I}$ crown height overlapped by $\bar{1}$. Crossbite (CB) referred to the buccolingual deviation of $\bar{M}$, measured as the proportion of the mesiolingual cusp displaced from the central occlusal fossa of $\bar{M}$. CB type was positive if $\bar{M}$ occluded buccally and negative if $\bar{M}$ occluded more lingually; CB extent was the absolute value. Buccal segment relation (BSR) referred to the sagittal deviation of the mesiobuccal cusp of $\bar{M}$ from alignment over the buccal groove of $\bar{M}$, expressed as a proportion of the cusp's size. BSR was also scored as type and extent, with mandibular retrognathism given a positive value. Both CB and BSR were scored on the side with maximum score. CB and BSR scores were arbitrarily incremented by from 0.2 to 0.4 when bilateral discrepancy was noted and when premolars and second molars were involved in the discrepancy. Rotation and displacement (R/D) was the sum of malaligned, rotated and crowded teeth. Anterior

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and post-canine teeth were scored separately and also summed to give total R/D. Treatment priority index (TPI), computed according to the method of Kelly and Harvey (1977), was a combined index used in this study merely to provide a convenient summary of the individual variables.

The approach we have adopted for the genetic analysis follows that of previous studies in US and Indian twins (Corruccini and Potter, 1980; Sharma and Corruccini 1986). *Several hidden assumptions implicit in the traditional twin model have been documented by Christian et al. (1974) and Kang et al. (1977).*

* Twin zygosity should not be associated with the mean of the trait under consideration (Christian and Norton 1977, Christian 1979). A modified t-test based on nested twin data has been recommended in which among-pair mean squares are used as the error term and the degrees of freedom are approximated (Christian et al. 1974; Christian 1979). Significant differences in mean values between MZ and DZ twins would reflect inherent biological differences associated with the twinning process (Christian and Norton, 1977).

* The standard twin model of Haseman and Elston (1970) partitions total variance into within -DZ(WDZ) and within -MZ(WMZ) mean squares. Christian et al. (1974) consider that total variance within zygosity must be equal for the model to hold. If there is evidence of heterogeneity of total variance, the environmental factors are postulated to be unequal for MZ and DZ twins. The arithmetic mean of among and within-mean squares (AMS and WMS) is then recommended to provide an unbiased estimate of twin genetic variance (Christian et al. 1974). These environmental factors may result from competitive or convergent influences that differ for the two twin types (Kempthorne and Osborne, 1961). To test heterogeneity of total variances, one-way analysis of variance is performed, first treating twin pairs as groups of two to provide among-pair and within-pair mean squares for MZ and DZ categories, then an F-test (Christian et al. 1974) compares $TMZ(AMZ + WMZ)$ and $TDZ(ADZ + WDZ)$. The larger value is used as the numerator of a two-tailed F-test, and the 0.2 probability level is used to control for type 2 error as the test is relatively insensitive to common variance in the zygosity.

* Genetic variance estimates will also be biased by inequality of environmental covariances of MZ and DZ twins (Christian 1979). If environmental covariance is relatively greater for MZ than for DZ twins ($C_{MZ} > C_{DZ}$), heritability will be exaggerated. To test this, Christian et al. (1975) propose an F test contrasting the within-pair and among-pair mean squares of DZ twins ($F = ADZ/WDZ$). If this ratio fails to appreciably exceed a value of

1, then the evidence for genetic variance rests solely in the MZ twins, and it is unlikely that any substantial proportion of the total variance is genetic (Christian et al. 1975; Christian 1979).

The classic genetic variance ratio (GVR) is calculated as $F = WDZ/WMZ$ and tested for significance, provided the data pass the above tests. If the F-test yields a significant result, a modified among-component ratio, $F_{ac} = (WDZ + AMZ)/(WMZ + ADZ)$, is used to provide an unbiased estimate of GVR. The rationale for this test is that AMZ/ADZ is likely to be just as valid and sensitive as WDZ/WMZ when total variance is heterogeneous (Christian et al. 1974).

We have presented basic statistics such as means, variances and intraclass correlation coefficients, r , for MZ and DZ twins. Three different estimates of heritability have also been calculated to quantify the proportion of total variance attributable to genetic influences. The within-pair heritability estimate h_{WP}^2 was computed as $h_{WP}^2 = 4(WDZ - WMZ)/(TDZ + TMZ)$ following Kang et al. (1978). The Holzinger heritability coefficient $h_H^2 = (r_{MZ} - r_{DZ})/(1 - r_{DZ})$ was also calculated as it has often been presented in previous twin analyses. In addition, the so-called path analysis model heritability estimate $h_t^2 = 2(r_{MZ} - r_{DZ})$ used recently by Lundström (1984), was also determined. Each of these estimates can theoretically range from zero to 1 (or 0-100%) reflecting the proportion of observed phenotypic variation due to genetic factors. However, negative values or values greater than unity are mathematically possible raising interpretative problems. Estimates derived from twin studies are referred to as broad heritabilities as the genetic influences may include additive, dominance and epistatic effects.

RESULTS

Table 1 gives basic descriptive statistics and values of the t' test for equality of mean values in MZ and DZ groups. Only one comparison, CB type, yielded a significant difference although this could well have been due to chance (Type 1 error). Furthermore, there did not appear to be any systematic differences in variances between zygosity. However, comparisons of the values of intraclass correlations were of some interest. Under a strictly polygenic mode of inheritance with additive genetic effects we would expect maximum values of 1.00 for MZ twins and 0.50 for DZ twins. The values of MZ twins were all considerably less than the theoretical maximum, emphasising that the occlusal traits are modified by cultural or environmental factors. Values of r for DZ twins were also generally less than the theoretical maximum, although several were quite close to 0.5 (for example OB, BSR, R/D) and TPI even exceeded this value.

Table 1. Descriptive statistics and t' tests for equality of MZ and DZ means

Occlusal traits	MZ twins(n=48pairs)			DZ twins(n=34pairs)			
	Mean	Variance	r	Mean	Variance	r	t'
OJ	2.96	1.88	0.75*	2.97	1.53	0.30*	-0.02
OB	0.42	0.04	0.70*	0.45	0.05	0.43*	-0.86
CB type	0.02	0.04	0.40*	0.12	0.09	0.04	-2.20*
CB extent	0.12	0.03	0.28*	0.18	0.07	0.02	-1.55
BSR type	0.22	0.22	0.69*	0.28	0.20	0.40*	-0.73
BSR extent	0.36	0.14	0.53*	0.36	0.15	0.41*	-0.10
PosR/D	0.83	0.94	0.54*	0.79	0.58	0.50*	0.23
AntR/D	2.82	2.61	0.60*	3.44	3.68	0.40*	-1.79
Total R/D	3.67	4.16	0.66*	4.26	5.12	0.49*	-1.39
TPI	3.31	6.30	0.71*	3.90	6.74	0.61*	-1.14

*p<0.05

r intraclass correlation

Table 2 Analysis of variance showing the among-pair mean squares (AMS) and within-pair mean squares (WMS) for MZ and DZ twins, the test of equality of total variance and the test to exclude $C_{MZ} > C_{DZ}$.

	MZ		DZ		Equality F'	$C_{MZ} > C_{DZ}$ $F = ADZ/WDZ$
	AMS	WMS	AMS	WMS		
OJ	3.310	0.471	1.998	1.084	1.23	1.84
OB	0.075	0.013	0.075	0.030	1.19	2.49
CB type	0.057	0.024	0.094	0.086	2.20 ¹	1.09 ²
CB extent	0.033	0.019	0.071	0.068	2.67 ¹	1.05 ²
BSR type	0.373	0.069	0.283	0.120	1.10	2.36
BSR extent	0.215	0.066	0.212	0.088	1.07	2.41
Pos R/D	1.454	0.438	0.882	0.294	1.61	3.00
Ant R/D	4.202	1.052	5.175	2.235	1.41	2.31
Total R/D	6.943	1.438	7.674	2.647	1.23	2.90
TPI	10.867	1.827	10.948	2.648	1.07	4.13

¹ DZ significantly more variable than MZ² $p > 0.10$ **Table 3** Estimates of genetic variance (GVR), heritabilities (h^2) and cultural inheritance (c^2) for occlusal traits in South Australian twins.

Occlusal traits	GVR	h^2_{wp}	h^2_H	h^2_c	c^2
OJ	0.61*	0.36*	0.65*	0.91*	-0.15
OB	0.02*	0.34*	0.47*	0.54*	0.16
CB type	No estimates as mean values differ between MZ and DZ				
CB extent	(0.01)	(0.24)	0.26	0.52	-0.24
BSR type	0.05*	0.24*	0.47*	0.57*	-0.11
BSR extent	0.02	0.15	0.20	0.23	0.29
Pos R/D	-0.14	-0.19	0.07	0.07	0.46
Ant R/D	1.18*	0.37*	0.34	0.41	0.20
Total R/D	1.21*	0.26*	0.33	0.34	0.32
TPI	0.82	0.12	0.26	0.20	0.51

GVR refers to within-pair variance, except for CB extent where among-component estimates of variance and heritability are given in parenthesis because of variance heterogeneity.

h^2_{wp} within-pair heritability estimate

h^2_H Holzinger heritability estimate

h^2_c intraclass heritability estimate

c^2 cultural inheritance calculated as $2r_{DZ} - r_{MZ}$

* $p < 0.05$

The results of analysis of variance showing among-pair mean squares (AMS) and within-pair mean squares (WMS) for MZ and DZ twins, the F test for equality of total variance, and the F test to check against environmental covariance are presented in Table 2. The F' test was significant for only one feature, CB, with DZ twins being significantly more variable than MZ twins. The ADZ/WDZ ratio was significant for all variables, with the exception of CB.

Genetic variance and heritability estimates are presented in Table 3. Five of the 10 occlusal variables (OJ, OB, BSR type, Ant R/D, Total R/D) displayed significant genetic variance according to the criteria of Christian et al. (1974). Within-pair heritability estimates for these traits ranged from 0.26 for Total R/D to 0.36 for OJ and 0.37 for Ant R/D.

DISCUSSION

Although it is generally agreed that a polygenic mode of inheritance is most likely to explain occlusal variation (Smith and Bailit, 1977), our findings indicate that environmental effects contribute more to observed

variability than early twin studies suggested. Significant genetic variance was noted for incisal overbite and overjet, sagittal molar relationship, and also rotations and displacements, particularly in anterior teeth. However, heritability estimates were generally low to moderate in magnitude, within-pair estimates for these traits averaging around 30%. Considering the problems of sampling and differences in methodology, our estimates are remarkably similar to those of Chung and Niswander (1975) and Lundström (1984). Furthermore, the trend noted by Harris and Smith (1980) in a non-western population for *genetic components of variance to be greatest for overjet, less for overbite and least for molar relationships* is confirmed in the present study.

Another interesting feature is the apparent lack of genetic determination for crossbite, a relationship in the coronal plane. This is consistent with morphometric studies that indicate greater transmissibility for body length than breadth measures (e.g. Byard et al. 1985; Devor et al. 1986). Indeed, in contrast with results from US and Indian twins where several occlusal traits displayed variance inequalities between zygosity (Corruccini and Potter, 1980; Sharma and Corruccini, 1986), crossbite was

the only variable in our study that failed to fulfil the assumptions of equality of mean values and variances.

The marked differences between heritability estimates for some of the occlusal traits (e.g. OJ), depending on their method of calculation, highlights the need for caution in interpreting these values. The path model estimate of 0.91 for OJ is undoubtedly inflated while the within-pair value of 0.36 provides a more conservative estimate. Indeed, Potter and Nance (1976) have argued that genetic variance ratios are generally more informative than heritability estimates in studies of human populations.

Researchers are becoming more cautious nowadays when extrapolating the results of twin studies to the general population. One must ask whether twins differ from singletons for the trait(s) under consideration. Boklage (1984) has provocatively stated on the basis of comparisons of dental crown size data that 'MZ twins, DZ twins, and singletons are not the same kind of people when the question is about the way their heads are built'. We intend to explore this question further in future studies, but at least in the present investigation there was little evidence of differences in distributional properties of occlusal data between MZ and DZ twins. Mean values of occlusal traits in MZ twins did not consistently exceed those in DZ twins as had been reported in Indians (Sharma and Corruccini, 1986). In fact, the trend was in the opposite direction, if anything. Only two instances of significant variance heterogeneity were noted (for CB type and extent), reflecting greater total DZ variability than MZ, again contrasting previous results in an Indian sample (Sharma and Corruccini, 1986). Many of the occlusal traits we have studied are inter-related and it is possible that multivariate approaches might disclose differences in distributional relationships between zygosity not evident in univariate analyses. In this regard, we have performed some preliminary discriminant function analyses. In contrast to the findings of Boklage (1984), they have not disclosed any statistically significant differences, although larger independent sample sizes are desirable for these types of approaches.

The question of whether the correlations noted between twins for occlusal traits reflect common genetic effects or common environmental influences is critical. Traditionally, the twin model assumes that environmental effects are similar for both MZ and DZ twins, and that greater similarity with MZ pairs compared with DZ pairs reflects genetic influences. Lundström (1984) and Lundström and McWilliam (1987) have calculated an estimate of cultural inheritance, according to the formula $c^2 = 2r_{DZ} - r_{MZ}$, to indicate the importance of familial similarity due to common familial environment rather than genetic effects. When this approach is adopted in our study, we find quite high values of c^2 for some traits, for example, posterior tooth rotations and displacements (0.46) and also TPI (0.51), pointing to important cultural factors (Table 3). However, the possibility that sampling effects may have inflated DZ correlations needs to be acknowledged. As we excluded any twin pairs where one or both members had a history of orthodontic treatment or permanent tooth loss, we effectively truncated the complete distribution of occlusal variation. Inclusion of a greater proportion of twin pairs with similar 'good' occlusions could have tended to inflate correlations, particularly in the DZ category where greater within-pair discrepancies might otherwise be expected. On the other hand, this unavoidable sampling bias would have inflated the ADZ/WDZ ratio, hiding environmental covariance from the $C_{MZ} < C_{DZ}$ test.

The twin model provides a valuable means of partitioning variability into causal components but a number of assumptions should be checked first before extrapolating results to the general population. As our sample sizes increase we intend to undertake more penetrating genetic analyses, for example, fitting models

that include parameters for individual environmental variation, additive genetic and dominance variation. These population-based quantitative approaches, together with more detailed longitudinal assessment of twin pairs where one or both members have received orthodontic treatment, should provide further insights into the nature and relative importance of the various determinants of occlusal variation.

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REFERENCES

- BOKLAGE, C.E. (1984)
Differences in protocols of craniofacial development related to twinning and zygosity
J. Craniofac. Genet. Devel. Biol. 4:151-169
- BYARD, P.J., POOSHA, D.V.R., SATYANARAYANA, M. and RAO, D.C. (1985)
Family resemblance for components of craniofacial size and shape
J. Craniofac. Genet. Devel. Biol. 5:229-238
- CHRISTIAN, J.C. (1979)
Testing twin means and estimating genetic variance. Basic methodology for the analysis of quantitative twin data
Acta Genet. Med. Gemellol. 28:35-40
- CHRISTIAN, J.C. and NORTON, J.A. (1977)
A proposed test of the difference between the means of monozygotic and dizygotic twins
Acta Genet. Med. Gemellol. 26:49-53
- CHRISTIAN, J.C., KANG, K.W. and NORTON, J.A. (1974)
Choice of an estimate of genetic variance from twin data
Am. J. Hum. Genet. 26:154-161
- CHRISTIAN, J.C., FEINLEIB, M. and NORTON, J.A. (1975)
Statistical analysis of genetic variance in twins
Am. J. Hum. Genet. 27:807
- CHUNG, C.S. and NISWANDER, J.D. (1975)
Genetic and epidemiologic studies of oral characteristics in Hawaii's schoolchildren v. Sibling correlations in occlusion traits
J. Dent. Res. 54:324-329
- CORRUCCINI, R.S. (1984)
An epidemiologic transition in dental occlusion in world populations
Am. J. Orthod. 86:419-426
- CORRUCCINI, R.S. and POTTER, R.H.Y. (1980)
Genetic analysis of occlusal variation in twins
Am. J. Orthod. 78:140-154
- DEVOR, E.J., MCGUE, M., CRAWFORD, M.H. and LIN, P.M. (1986)
Transmissible and non-transmissible components of anthropometric variation in the Alexanderwohl Mennonites:
1. Description and familial correlations
Am. J. Phys. Anthropol. 69:71-82
- HARRIS, E.F. and SMITH, R.J. (1980)
A study of occlusion and arch widths in families
Am. J. Orthod. 78:155-163
- HASEMAN, J.K. and ELSTON, R.C. (1970)
The estimation of genetic variance from twin data
Behav. Genet. 1:111-119
- KANG, K.W., COREY, L.A., EVANS, M.M., CHRISTIAN, J.C. and NORTON, J.A. (1977)
Dominance and environmental variances, their effects on heritabilities estimated from twin data
Hum. Hered. 27:9-21
- KANG, K.W., CHRISTIAN, J.C. and NORTON, J.A. (1978)
Heritability estimates from twin studies. 1. Formulae of heritability estimates
Acta Genet. Med. Gemellol. 27:39-44
- KELLY, J.E. and HARVEY, C.R. (1977)
An assessment of the occlusion of the teeth of youths 12-17 years
Vital and Health Statistics, Series 11, No. 162, US Public Health Service, Washington
- KEMPTHORNE, O. and OSBORNE, R.H. (1961)
The interpretation of twin data
Am. J. Hum. Genet. 13:320-339
- LUNDSTRÖM, A. (1984)
Tooth Size and Occlusion in Twins
Karger, Basle
- LUNDSTRÖM, A. (1984)
Nature versus nurture in dento-facial variation
Europ. J. Orthod. 6:77-91
- LUNDSTRÖM, A. and MCWILLIAM, J.S. (1987)
A comparison of vertical and horizontal cephalometric variables with regard to heritability
Europ. J. Orthod. 9:104-108
- POTTER, R.H. and NANCE, W.E. (1976)
A twin study of dental dimension. 1. Discordance, asymmetry and mirror imagery
Am. J. Phys. Anthropol. 44:391-395
- POTTER, R.H.Y., CORRUCCINI, R.S. and GREEN, L.J. (1981)
Variance of occlusion traits in twins
J. Craniofac. Genet. Devel. Biol. 1:217-227
- PROFFIT, W.R. (1986)
On the aetiology of malocclusion
Brit. J. Orthod. 13:1-11

- SHARMA, K. and CORRUCINI, R. (1986)
Genetic basis of dental occlusal variations in northwest Indian twins
Europ. J. Orthod. 8:91-97
- SMITH, R.J. and BAILLIT, H.L. (1977)
Problems and methods in research on the genetics of dental occlusion
Angle Orthod. 47:65-77
- STEIN, K.F., KELLEY, T.J. and WOOD, E. (1956)
Influence of heredity in the etiology of malocclusion
Am. J. Orthod. 42:125-141

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