Heritability of craniometric and occlusal variables: A longitudinal sib analysis

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There has long been interest in the inheritance of malocclusion, but few studies have distinguished between skeletal (craniometric) variables and occlusal, tooth-based variables (e.g., anterior irregularity, rotations, displacements). This study was based on serial assessments of untreated persons in 30 sibships from 4 years (full deciduous dentition) to 20 years of age (full permanent dentition) in the Bolton-Brush Growth Studies of Ohio. Results define a clear dichotomy: craniometric variables (k = 29) typically show significant additive components of variance; correlations increase from age 4 to age 20; and correlations average 0.43 at adulthood. Tooth-based variables of position and relationship (k = 21) reach significance only occasionally; correlations decrease with age to the extent that few variables for subjects at age 20 have a correlation significantly different from zero. In contrast to craniometric variables, which have high heritabilities, almost all of the occlusal variability is acquired rather than inherited. (Am J Orthod Dentofac Orthop 1991;99: 258-68.)

Malocclusion is a major developmental problem in this and other industrialized countries. Kelly and Harvey[1] report that only about 9% of American youths aged 12 to 17 years have virtually classic normal occlusion and that orthodontic treatment would measurably improve the occlusion of 55% of the adolescent population. This high prevalence is a major health care concern, and it is only natural to be interested in the causes of malocclusion. The issue is particularly relevant when viewed from the perspective that prehistoric and contemporary preindustrialized peoples have substantially lower prevalences of dental, skeletal, and skeletodental malocclusions.[2-4]

Contemporary clinical opinion emphasizes the role of heredity as a cause of malocclusion. In craniometric and cephalometric studies of familial similarities, a good deal of evidence supports the contention that facial form is largely a product of the person's genotype.[5-10] But it does not necessarily follow that tooth-based malocclusions is also inherited. Indeed, there is a confusion in the literature between the causes of bone- and tooth-based malocclusion (Fig. 1).[11,12] While many types of malocclusions involve skeletal disharmony and problems with tooth position and tooth-to-tooth relationships, few researchers have distinguished between these types of condition, which may have quite different causes.

In a studied review of the literature available through the 1970s, Smith and Bailit[13] concluded that heredity played a far greater role than the environment in the development of "malocclusions." In this past decade, however, several researchers have questioned this conventional wisdom. Perhaps foremost has been the methodologic stride in clarification of the several implicit sources of error in the classic studies of twins.[14-17] Failure to account for unequal means and unequal variances in mono- and dizygotic twin samples introduces serious biases in most of the work to date.[18-21]

In addition, research during the past few years,[22-25] based on both twin and sib analyses, has questioned the facile lumping of bone- and tooth-based variables into a common category of "malocclusion." Instead, the role of heredity in the expression of these two sorts of skeletodental variable needs to be assessed individually.[17,22,26]

The intent of this study was to assess the relative contributions of heredity to the phenotypic expression of a variety of craniofacial skeletal dimensions and tooth-based occlusal variables. We were concerned, specifically, with testing for differences in sibling similarities among these kinds of variable. In addition, we have used longitudinal growth information to monitor the changing effects, with age, of heredity and environment on skeletodental variables.

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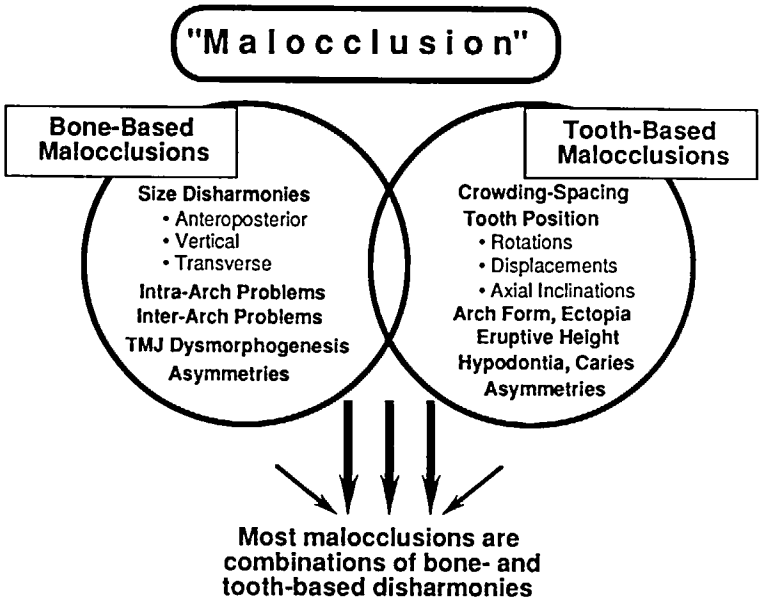

Fig. 1. Schematic representation of the interrelationships between bone- and tooth-based sources of malocclusion (considerably modified from Case[11] and Ackerman and Proffit[12]). Some individuals have predominantly one or the other source of "malocclusion"; the common occurrence of cases with both (e.g., a skeletal Class II with blocked-out canines and lower incisor crowding) often confuses the issue of causes, since co-occurrence should not imply a single or even a similar cause for bony- versus tooth-based disharmonies. A wide variety of treatment concerns (e.g., overjet, overbite, molar relationship) may be wholly dental or skeletal or, often, some composite of each.

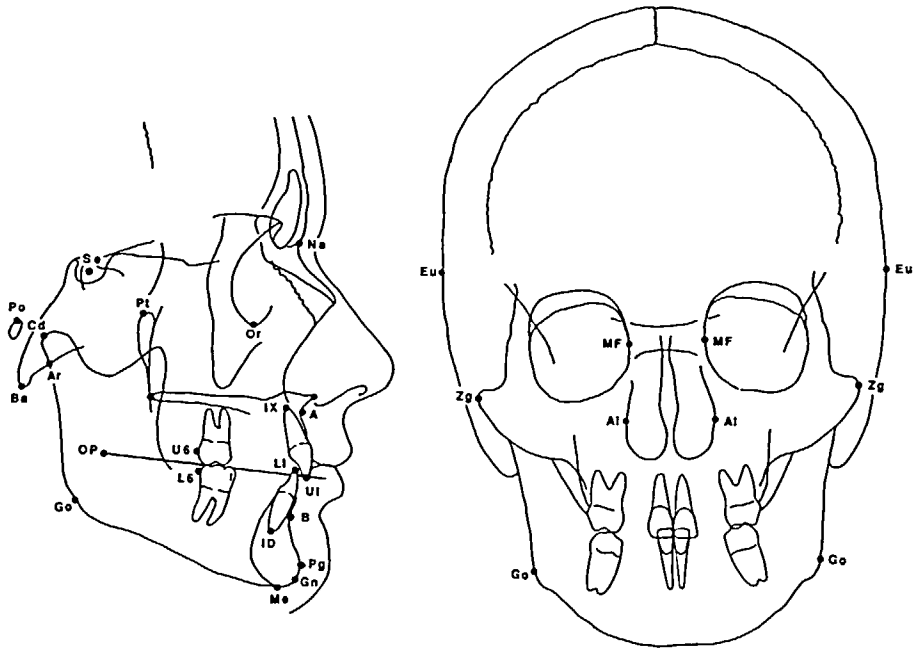

Fig. 2. Identification of the 23 landmarks traced from the lateral cephalogram and the five paired landmarks recorded from the posteroanterior film.

Table I. Measurements and definitions of lateral and frontal cephalometric variables, the arch dimensions, and occlusal variables*

Craniofacial parameters

1. Sella-gnathion distance (mm)
2. Nasion-menton distance (mm)
3. Sella-nasion distance (mm)
4. Sella-A point distance (mm)
5. Sella-B point distance (mm)
6. Sella-posterior nasal spine (PNS) distance (mm)
7. Sella-gonion distance (mm)
8. Nasion-anterior nasal spine (ANS) distance (mm)
9. Sella-nasion-A point (SNA) angle (degrees)
10. Sella-nasion-B point (SNB) angle (degrees)
11. A point-nasion-B point (ANB) angle (degrees)
12. Sella-nasion-palatal plane (S-N to ANS-PNS) angle (degrees)
13. Gonion-PNS distance (mm)
14. Articulare-B point distance (mm)
15. Articulare-pogonion distance (mm)
16. Gonion-pogonion distance (mm)
17. Sella-nasion to mandibular plane (Go-Me) angle (degrees)
18. Frankfort horizontal (Po-Or) to mandibular plane angle (degrees)
19. AO-BO discrepancy (mm): The distance along Downs' occlusal plance between the perpendicular projections of points A and B[29]
20. Bi-euryon breadth (mm): Left and right euryon are the two points on opposite sides of the calvaria viewed in *norma frontalis* that yield the greatest breadth measured parallel to the orbital plane[30]
21. Bi-maxillofrontale breadth (mm): Maxillofrontale is the point of intersection of the anterior lacrimal crest (medial edge of the orbit), or the crest extended, with the frontomaxillary suture
22. Bi-zygion breadth (mm): Zygion is the most lateral point on the zygomatic arch when viewed in norma frontalis[30]
23. Bi-alare breadth (mm): Alare is the most lateral point on the nasal aperture taken parallel to the nasal height[30]
24. Bi-gonion breadth (mm): Distance between the lateral borders of the left and right gonial margins on the frontal cephalogram
25. Total height index: Nasion-menton distance divided by sella-gonion distance
26. Anterior height index: Nasion-ANS distance divided by nasion-menton distance
27. Total depth index: Sella-A point distance divided by articulare-pogonion distance
28. Upper depth index: Sella-nasion distance divided by sella-A point distance
29. Facial width index: Bi-zygion width divided by bi-gonion width

*Traits were categorized as measures of craniofacial (skeletal), arch (essentially alveolar-bone based), and occlusal (tooth position and relation) parameters. Of course, some involve more than one of these categories (e.g. overjet, BSR), but a decision was made *a priori* about the major source of influence. When bilateral asymmetries occurred (e.g., overjet, overbite), the more extreme condition was recorded.

MATERIALS AND METHODS

Sample selections

Records were obtained from the Bolton-Brush Study collections housed at Case Western Reserve University in Cleveland, Ohio. Participants had been chosen because they were well nourished, well developed, in good health, and progressing well in school[27]; they also tended to be of middle to high socioeconomic status. Annual radiographic records and dental models are available for most participants from birth through approximately 20 years of age.

Subjects were selected according to three criteria: (1) there was at least one same-sex sibling who had also participated in the Bolton-Brush study, (2) records for the three ages were available for evaluation, and (3) neither sibling had received orthodontic treatment. There was a normal range of types of malocclusion in this series, with both Class II and Class III sagittal molar relationships, but extreme cases were eliminated insofar as the subjects (a) had not been enrolled in the study of "healthy children" or (b) had received orthodontic treatment. Individual subjects were evaluated at three ages: 4 years (full complement of deciduous teeth), 14 years (early permanent dentition), and 20 years of age (early adulthood). Records were obtained for 16 male sibships (n = 34) and 14 female sibships (n = 31). The longitudinal records had invariably been taken within a few days of birth, so there was no need to correct for age variation.

The exclusion of extreme malocclusions, a result

Table I. cont'd

Arch parameters

30. Maxillary 3-3 width: Intercanine width measured as the maximum distance at the buccal surfaces of the canines, either deciduous or permanent[31,32]
31. Mandibular 3-3 width
32. Maxillary length: Linear distance (chord) from the interincisal (1-1) midline to the distobuccal aspect of the first molar (deciduous or permanent)[32]
33. Mandibular length
34. Maxillary shape index: Maxillary intercanine width divided by maxillary arch length
35. Mandibular shape index
36. Incisor overjet: The horizontal distance (mm) from the labial surface of the maxillary central incisor to the surface of the lower central incisor measured parallel to the occlusal plane[33]

Occlusal parameters

37. Interincisal angle: Measured from the lateral cephalogram, this is the posterior angle formed at the intersection of the long axes of the upper and lower central incisors[34]
38. Incisor-mandibular plane angle (IMPA): The posterior-superior angle formed by the long axis of the lower central incisor and the mandibular plane (Go-Me) as seen on the lateral cephalogram[34,35]
39. Incisor overbite: The amount of vertical overlap (mm) of the maxillary and mandibular central incisors measured perpendicular to the occlusal plane; openbite (apertognathia) was assigned a negative value[33]
40. Buccal segment relationship (BSR): The anteroposterior distance (mm) between the mesiobuccal cusp tip of the maxillary first molar and the buccal groove of the mandibular first molar measured parallel with the occlusal plane[23,32]
41. Maxillary crowding: A qualitative, ordinal scale with seven categories ranging from severe crowding (−3) in the anterior (incisor-canine) region to ideal occlusion (O) to severe spacing (+3) was used; categories were defined to approximate equal distances between intervals[23,32]
42. Mandibular crowding
43. Maxillary incisor irregularity: Sum (mm) of the five distances between the anatomic contacts from the mesial aspect of the left canine throught the mesial aspect of the right canine[36,37]
44. Mandibular incisor irregularity
45. Crossbites: The count of the number of maxillary premolars and molars in ligual or (rarely) buccal crossbite, summed across the left and right quadrants but excluding third molars[1,22]
46. Maxillary rotations: The weighted count of premolars and molars rotated out of presumed ideal alignment, with teeth rotated more than 15° up to 45° given a weight of 1 and teeth rotated more than 45° given a weight of 2[1,38]
47. Mandibular rotations
48. Maxillary displacements: The weighted count of premolars and molars displaced out of ideal alignment, with teeth displaced buccally or lingually up to 2 mm, given a weight of 1 and teeth displaced more than 2 mm given a weight of 2[1,38]
49. Mandibular displacements
50. Summed rotations and displacements: Summation of the weighted rotation and displacement scores for the four quadrants

of orthodontic treatment, introduced a potential bias, since it also reduces intra-group variation. On the other hand, it reduces intra-sibship variation, so that the ratios of variability both within groups and among groups, on which transmissibility estimates are based, probably were not affected substantially. We have subsequently addressed this issue by focusing on sibling pairs with overt malocclusion[28] and have confirmed that the present phenotypic distributions yield comparable results.

Cephalometrics

Standardized lateral and frontal cephalograms were traced (Fig. 2), and the landmarks were digitized with the aid of a microcomputer. In all, 29 measurements were computed from each pair of radiographs (Table I): 18 linear, 6 angular, and 5 proportional. The distances from each subject's midline of the base to film (ML distance) on the lateral radiograph and from the center of head to film on the posterior film (P+ distance) were used to correct each variable for magnification.[39]

Cast analysis

Twenty-two variables of occlusion were measured on each of the full-mouth dental casts for each sibling (Table I). These traits are broadly divisible into two categories, those that reflect development of the arch (e.g., length, width) and those that quantify tooth position (e.g., incisor irregularity, tooth rotations, and displacements within the arch form).

Statistical analysis

Sex-specific and pooled intraclass correlations (r_i) and heritability estimates (h^2) were computed for each variable with the use of intraclass correlation from

Table II. Heritability estimates derived from intraclass correlation coefficients

Parameter	Age 4				Age 14				Age 20			
	n	h^2	*SE*	*F ratio*	*n*	h^2	*SE*	*F ratio*	*n*	h^2	*SE*	*F ratio*
Craniofacial parameters												
Sella-gnathion	45	0.62	0.37	2.0*	53	1.45	0.18	6.6*	47	1.61	0.15	9.3*
Nasion-menton	45	0.12	0.39	1.1	53	1.29	0.23	4.9*	47	1.56	0.16	8.2*
Sella-nasion	45	0.50	0.38	1.8	53	0.62	0.34	2.0*	47	1.00	0.31	3.0*
Sella-A point	45	1.39	0.22	6.1*	53	0.67	0.34	2.1*	47	1.16	0.28	3.8*
Sella-B point	45	0.40	0.38	1.6	53	1.23	0.24	4.4*	47	1.28	0.25	4.6*
Sella-PNS	45	0.57	0.37	1.9*	53	0.98	0.29	3.0*	47	1.46	0.19	6.5*
Sella-gonion	45	0.65	0.36	2.1*	53	0.94	0.30	2.9*	47	1.35	0.22	5.3*
Nasion-ANS	45	0.94	0.32	3.0*	53	1.33	0.22	5.2*	47	0.97	0.32	2.9*
SNA angle	45	0.55	0.37	1.8	53	0.68	0.34	2.1*	47	0.27	0.40	1.3
SNB angle	45	0.62	0.37	2.0*	53	0.93	0.30	2.8*	47	0.37	0.40	1.5
ANB angle	45	0.84	0.34	2.6*	55	0.61	0.34	1.9*	47	0.27	0.40	1.3
SN-PP angle	45	0.82	0.34	2.6*	55	1.22	0.24	4.3*	47	0.21	0.39	1.5
Gonion-PNS	45	0.80	0.34	2.5*	53	0.57	0.35	1.8	47	0.76	0.35	2.3*
Articulare-B point	45	0.51	0.38	1.8	53	1.08	0.27	3.5*	47	1.16	0.27	3.8*
Articulare-pogonion	45	0.75	0.35	2.3*	53	1.20	0.25	4.2*	47	1.36	0.22	5.4*
Gonion-pogonion	45	0.22	0.39	1.3	53	0.67	0.34	2.1*	47	0.65	0.37	2.0*
Sella-nasion MP angle	45	0.60	0.37	2.0*	53	0.21	0.37	1.3	47	0.35	0.40	1.4
FH MP angle	45	0.60	0.37	2.0*	53	0.62	0.34	2.0*	47	0.49	0.39	1.7
AO-BO discrepancy	45	0.52	0.38	1.8	53	0.62	0.35	1.9*	47	−0.06	0.41	0.9
Bi-euryon	43	0.15	0.40	1.2	55	0.52	0.35	1.7	47	−0.35	0.39	0.7
Bi-maxillofrontale	43	0.81	0.35	2.5*	55	1.06	0.27	3.4*	47	0.99	0.31	3.0*
Bi-zygion	43	0.68	0.37	2.2*	55	1.13	0.26	3.8*	47	1.38	0.22	5.5*
Bi-alare	43	0.86	0.34	2.7*	55	1.05	0.27	3.3*	47	0.85	0.34	2.5*
Bi-gonion	43	0.81	0.35	2.5*	55	0.97	0.29	3.0*	47	0.91	0.33	2.7*
Total height index	45	0.67	0.36	2.1*	55	0.22	0.36	1.3	47	0.46	0.39	1.6
Anterior height index	45	1.19	0.27	4.3*	55	1.33	0.21	5.2*	47	0.83	0.34	2.5*
Total depth index	45	0.18	0.39	1.2	55	0.55	0.35	1.8	47	0.86	0.34	2.5*
Upper depth index	45	0.32	0.39	1.4	55	0.48	0.35	1.7	47	0.12	0.41	1.1
Width index	43	0.33	0.39	1.4	57	0.75	0.32	2.3*	47	1.01	0.31	3.1*

The number of siblings (n) varies by availability of measurable records; standard errors are for h^2, not r_i. F ratios are the results of tests for significant intersibship components of variation. Arcade abbreviations are maxilla (MX) and mandible (MD).
*$p < 0.05$.
†Left and right sides combined.
‡Trait is invariant, so h^2 could not be calculated.

a model II analysis of variance.[40,41] Heritability for siblings was defined as twice the intraclass correlation.[42-44] The standard error of each h^2 estimate was calculated from the formula in Swiger et al.[45] and Becker.[46]

All analyses were originally performed on the two sexes separately.[47] Tests for sexual dimorphism at each age were negative, which agrees with earlier findings.[23,48] To conserve space, only the data for the pooled sample are presented here.

RESULTS

Table II presents the heritability estimates and other information by age for each of the 50 variables. The theoretical upper limit of the genetic contribution for a first-degree relative is a correlation of 0.5, but, because of sampling fluctuation and environmental covariation (enhanced acquired similarity), correlations can exceed 0.5 and, indeed, have an upper boundary greater than 1.0.

CRANIOFACIAL VERSUS OCCLUSAL VARIABLES

Inspection of the individual scores in Table II suggests that craniofacial variables tend to yield higher h^2 estimates than arch size or occlusal parameters. To assess this impression statistically, Kruskal-Wallis one-way analysis of variance tests (H) were computed for differences in h^2 estimates by group.[49]

For subjects at age 4, H was 9.3 (df = 2), which is highly significant ($p < 0.001$; H is distributed as X^2). The palatal variables have higher average h^2 estimates than the craniofacial variables, which in turn

Table II. cont'd

Parameter	Age 4				Age 14				Age 20			
	n	h^2	*SE*	*F ratio*	*n*	h^2	*SE*	*F ratio*	*n*	h^2	*SE*	*F ratio*
Arch parameters												
MX 3-3 width	42	0.79	0.36	2.4*	54	0.71	0.33	2.2*	43	0.05	0.43	1.1
MD 3-3 width	40	0.94	0.34	3.0*	58	0.45	0.34	1.6	43	−0.20	0.42	0.8
MX length	42	1.34	0.24	5.5*	58	−0.52	0.31	0.6	43	−0.01	0.43	1.0
MD length	42	0.91	0.34	2.8*	56	0.22	0.36	1.3	43	−0.25	0.42	0.8
MX shape index	42	1.09	0.30	3.6*	54	−0.01	0.36	1.0	43	0.13	0.43	1.1
MD shape index	40	.58	40	1.9	56	0.62	0.33	2.0*	43	−0.49	0.40	0.6
Incisor overjet	42	−0.20	0.39	0.8	58	0.46	0.34	1.6	43	0.43	0.41	1.6
Occlusal parameters												
Interincisal angle	45	0.29	0.39	1.4	53	0.10	0.37	1.1	47	0.10	0.41	1.1
IMPA	45	0.45	0.38	1.7	53	0.24	0.37	1.3	47	0.76	0.35	2.2*
Incisor overbite	42	0.61	0.38	2.0	58	−0.02	0.35	1.0	43	0.19	0.42	1.2
Buccal segment relation†	88	0.54	0.23	2.5*	122	0.34	0.19	1.8*	92	0.24	0.21	1.5
MX crowding	42	0.64	0.38	2.0*	58	0.53	0.34	1.8	43	0.11	0.43	1.1
MD crowding	42	0.66	0.38	2.1*	58	0.85	0.30	2.6*	43	0.63	0.39	1.9
MX incisor-irregularity	40	0.53	0.40	1.8	58	−0.13	0.34	0.9	43	0.24	0.42	1.3
MD incisor-irregularity	42	0.66	0.38	2.1*	58	0.20	0.35	1.2	43	0.46	0.41	1.6
Posterior crossbites	42	0.14	0.41	1.2	58	−0.17	0.34	0.8	43	−0.05	0.43	0.9
MX rotations	‡	—	—	—	58	−0.15	0.34	0.8	43	−0.02	0.43	1.0
MD rotations	42	0.09	0.41	1.1	58	−0.48	0.31	0.6	43	−0.04	0.43	1.0
MX displacements	—	—	—	—	58	−0.17	0.34	0.8	43	−0.16	0.42	0.8
MD displacements	—	—	—	—	58	0.72	0.32	2.2*	43	0.66	0.38	2.0*
Summed rotations and displacements	42	0.09	0.41	1.1	58	−0.18	0.34	0.8	43	0.00	0.43	1.0

have higher estimates than the occlusal variables. Indeed, each of the three types of variable is statistically distinct from the other two.

The same test on subjects at age 14 yielded an H of 23.1 (2 df; $p < 0.001$). For this age group, the palatal variables have reduced heritability so that the single source of significance is that the craniofacial variables are higher, on average, than the palatal and occlusal variables. The occlusal variables continue to show very low heritabilities and, with growth from full deciduous dentition at age 4 to early permanent dentition at age 14, there is a decrease in the arch parameter heritability estimates.

For subjects at age 20, H was again significant at 18.7 ($p < 0.001$). As at age 14, the craniofacial variables have significantly higher average heritability than the arch dimensions, which in turn are not statistically distinguishable from the occlusal variables (craniofacial > palatal = occlusal).

AGE TRENDS

A complementary issue is whether the estimates reflect systematic changes with age. The Friedman two-way analysis of variance[49,50] was used to test for differences in mean h^2 across the ages of 4, 14, and 20 years.

Table III. Results of Mann-Whitney U tests for differences in h^2 estimates between vertically and horizontally oriented craniofacial parameters

Age	U	p Value
4	19	>0.60
14	9	>0.10
20	10	>0.10

Seven vertical dimensions:
1. Sella-gnathion
2. Nasion-menton
3. Sella-PNS
4. Sella-gonion
5. Sella-B point
6. Nasion-ANS
7. Gonion-PNS

Five horizontal dimensions:
1. Articulare-pogonion
2. Articulare-B point
3. Sella-A point
4. Sella-nasion
5. Gonion-pogonion

For the 29 craniofacial variables, the χ^2 test statistic is 6.3 ($p < 0.05$). This significance is attributable to the substantive increase from 4 to 14 years. There is no significant difference between ages 14 and 20, although the ranks are slightly higher for some traits at age 20.

For the seven palatal variables, χ^2 is 4.5 (2 df; $p > 0.20$), indicating no systematic change with age.

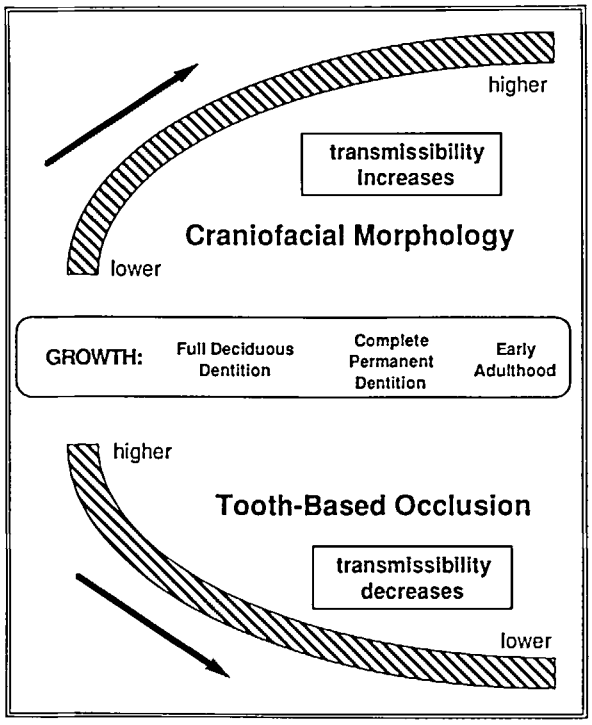

Fig. 3. The conceptual model derived from the present analyses. Trends across the three ages examined (4, 14, 20 yr) are in opposite directions for craniofacial variables (k = 29) and arch size and tooth-based measures of occlusion (k = 21). In both groups the significant changes occur between development of the full deciduous dentition, monitored at 4 years, and the early permanent dentition, at 14 years, with plateauing thereafter.

For the 14 occlusal variables, χ^2 is 12.0 (2 df; $p < 0.01$). The nature of this difference is that the h^2 estimates are significantly higher at age 4, with a significant decrease to age 14 and maintenance (nonsignificant change) from the ages of 14 to 20 years.

The palatal and occlusal variables were pooled, and the resultant χ^2 is 14.1 ($p < 0.01$), with the nature of the trend being identical to that for the occlusal variables alone.

VERTICAL CRANIOFACIAL VARIABLES

It has been suggested that vertically oriented craniofacial variables are more tightly controlled by the genotype than horizontally based dimensions.[19,51-53] To test this, the 12 variables that were either horizontal or vertical in their primary orientation were identified (Table III). Results of a Mann-Whitney U test[49] disclosed no significant difference at any of the three ages.

DISCUSSION

When genetic and environmental contributions are partitioned, the total genotypic contribution to the phenotypic variation is termed heritability in the broad sense. This variance can, in turn, be partitioned into contributions from individual alleles (additive variance), from pairs of homologous alleles at a locus (dominance variance), from combinations of nonhomologous loci (epistatic variance), and so forth.[43,54] In contrast, the potentially smaller proportion of phenotypic variance that can be attributed to additive genetic variance is called heritability in the narrow sense.[55] It is this estimate that is calculated when siblings, including twins, are compared; human mating patterns generally preclude finer discrimination of the sources of genetic variation, although intergenerational data can clarify several issues.[56,57]

Sibling correlations tend to overestimate the additive genetic component because they incorporate any dominance effect and all acquired similarities that result from shared environments. The influence of shared environments, what Garn et al.[58] have termed the "cohabitational effect," has gained increasing relevance over the past few years as more data become available.[59-62] The key issue is that cohabitational effects cause family members to appear more alike than they would appear simply because of sharing half their genes in common by descent. This is especially true in the case of siblings because of the added cohort effect.[63] Siblings (and especially twins in this culture) share the same maternal environment and very similar peri- and postnatal conditions, including a number of issues relevant to skeletodental development (e.g., dietary preferences, manner of food preparation, socioeconomic status, and patterns of energy expenditure and childhood illnesses). Simply living together, let alone growing and developing in the same household, can have far-reaching influences on phenotypic similarities; generally, these factors enhance phenotypic correlations. These observations also interject a cautionary note: the perception that siblings "look alike" is no proof that the cause is genetic; the similarities could as well be due to developmental convergences that result from the siblings being raised in similar environments.

Craniofacial versus occlusal variables

Numerous studies have examined the genetic contribution to craniofacial similarities among family members,[6,7,64-66] while others have similarly assessed occlusal variation.[17,22,23,26] Few studies have simultaneously determined h^2 for craniofacial and occlusal variables.

The fundamental conclusion to be drawn from the present study centers on the dichotomy in genetic control; the distinction is between craniofacial, dimensions with moderate h^2 estimates on the one hand and occlusal, tooth-based parameters with much lower h^2 values on the other. This difference can be synthesized as shown in Table IV, where median h^2 estimates from Table II are listed by type and age.

The median h^2 estimates start close to one another in the full deciduous dentition but then diverge substantially so that, by early adolescence and on into young adulthood, the craniofacial (bone-based) variables have large h^2 values, while measures of tooth relationships and alveolar-bone growth (arch size) diminish to essentially zero (Fig. 3).

The finding that craniometric dimensions have moderate to high similarities among siblings agrees well with those of prior studies.[6,64-67] In addition, at least two studies have assessed longitudinal cephalometric data and noted age effects on familial correlations consistent with those reported here. Byard et al.[68] and Kohn[69] have each presented sibling correlations for craniofacial measurements from lateral cephalograms. Correlations generally were lowest during childhood and increased as adulthood was approached.

The finding that occlusal variables have low familial correlations also is adumbrated by previous work. Nakasima et al.[70] computed parent-offspring correlations in children with Class II and Class III malocclusions and found the correlations to be significantly higher for skeletal variables than for dental variables (interincisal angle, overbite, and overjet) in both malocclusion groups. Corruccini et al.[17,22,71] and Harris and Smith[23] compared measures of tooth position with measures of arch dimensions and found that the occlusal variables, per se, had lower familial correlations. The conclusion of Corruccini et al.[26] is typical of several analyses: "We do not feel that any meaningful proportion of the occlusal variation . . . can be ascribed to genetic variance."

It merits noting in this regard that many tooth-based measures (Table I) share the combined variations of tooth position and basal and alveolar bone development.[72] Simply from a conceptual perspective, since teeth are positioned by the bones supporting them, their relationships (as in overjet, overbite, molar relationship) cannot be less variable than the supporting structures and, predictably, they will vary because of their own inexactitudes in formation as well as those of the basilar structures. Of course, this variability may be offset by dental compensations,[73] but these accommodations in tooth position and angulation, while reducing such traits as overjet and overbite, will *increase* tooth-based variability elsewhere (e.g., IMPA, interincisal angle, incisor irregularity).

Table IV. Median h^2 estimates (from Table II) by type and age

Age (yr)	*Craniofacial variables*	*Arch and occlusal variables*
4	0.6	0.5
14	0.9	0.2
20	0.9	0.1

One key source of confusion in discussing the causes of malocclusion has been the tendency to combine bone- and tooth-based types of malocclusion. As noted by Smith and Bailit,[13] "most studies have considered malocclusion to be largely synonymous with Angle's classification." In addition, there are numerous instances in which craniofacial (cephalometric) variables were assessed, but sweeping conclusions were drawn about "malocclusions" *(sensu latto)*. Thus, for example, Stein, Kelley, and Wood[64] computed interclass correlations among relatives for cephalometric angles and concluded that "malocclusions" were inherited; they extrapolated from facial dimensions to the entire range of malocclusion (Fig. 1). In a similar vein, virtually all reviews on the causes of "malocclusion" focus almost exclusively on skeletal disharmonies.[74-77]

Causes of malocclusion

Most texts on orthodontics deal in some fashion with the causes of malocclusions, but most listings fail to account for the preponderance of cases. Obvious causes include chromosomal and genetic disorders (and syndromes), trauma, caries, and such habits as thumb sucking, but, even collectively, these identifiable causes fall short of accounting for the high prevalence of malocclusion in industrialized countries[1,76] or the rapid increase in prevalence over the past few generations.[48] Other ideas once in vogue, such as radical outcrossing,[11,78] failed to account for the realities of genetics or of history.[79-81] Two sources of malocclusion that (a) can potentially account for large portions of the cases seen in contemporary populations and (b) have been shown to affect both craniofacial and occlusal components of malocclusion are the increased frequency of chronic mouthbreathing in westernized populations[81,82] and reduced masticatory stress, especially during formation of the dentition.[83-85] Singly and together, these constitute changes that researchers have termed an epidemiologic transition, that is, a shift in which morbidity from acute

and infectious diseases to predominantly chronic, degenerative, and man-made diseases.[86-89] Populations in westernized, industrialized countries have already experienced this transition (as exemplified by the increased prevalence of cancers, circulatory diseases, and hypertension), whereas the trends are ongoing in less westernized cultures.[90,91]

Studies by Hunt,[2] Lombardi and Bailit,[92] and Corruccini and Whitley[3] cite several instances of rapid transition in the prevalence of malocclusion that coincides with dietary changes and other changes in life-style. Subsequently, Corruccini[48] has thoughtfully reviewed a cross-cultural spectrum of studies, each consisting of at least two components. One component is preindustrialized (less-industrialized) and the other is influenced by a more contemporary diet that requires less masticatory stress. The consistent finding was an epidemiologic transition to higher frequencies of malocclusion, and the rapidity of the transition was proportional to the rate of urbanization. Oral influences readily associated with urbanization include caries, nasorespiratory obstructions, nutritional and growth differences, premature deciduous exfoliation, and dietary consistency.

The association between dietary consistency (masticatory function) and occlusal development is an important area of investigation. The chewing stress–stimulation model, which holds that occlusal variation is a major factor in jaw-to-tooth size discrepancies, provides one explanation for the occlusal disharmonies seen in urbanized modern populations,[48] and this explanation is substantiated by controlled experiments.[83-85,93]

In overview, this work reveals a fundamental dichotomy in the genetic contribution to the development of malocclusions: Craniometric measures have comparatively high heritabilities, while occlusal variables of tooth position have low genetic contributions that, generally, are not significantly different from zero. This finding contradicts the often cited results of Lundström[18] and those of Borass et al.[94] but is in close agreement with studies that account overtly for various sources of bias in the analyses.[16,17,22] Malocclusion, defined in the strict sense as tooth malpositions, is essentially an *acquired* condition. These results highlight the need to explore further the range of underlying maternal, "cohabitational," and other environmental *causes* of occlusal variation.

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