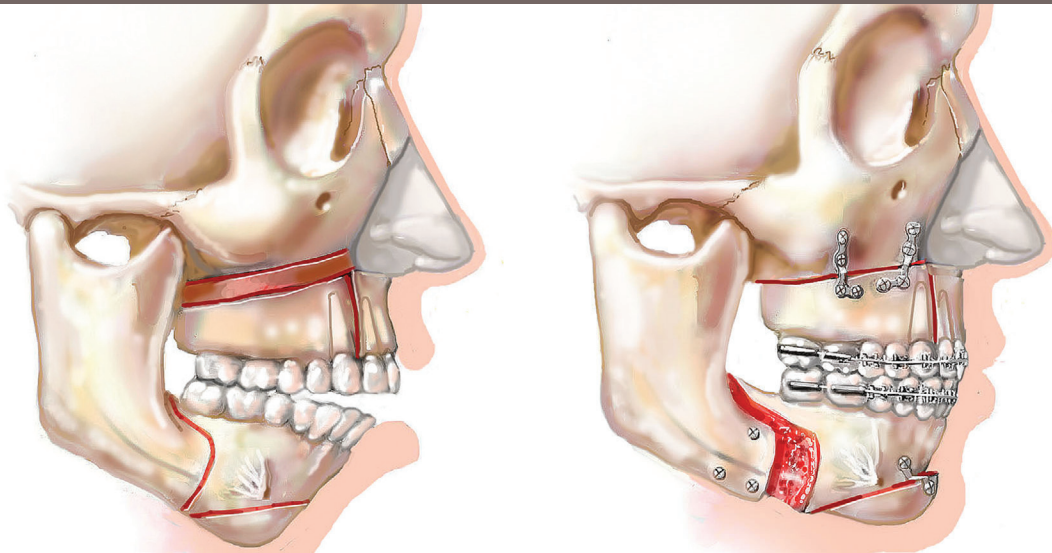


JEFFREY C. POSNICK, DMD, MD

ORTHOGNATHIC
SURGERY:
PRINCIPLES & PRACTICE

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SURGERY:
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To my wife, best friend and Soul mate Patti, and our two sons,
Joshua and David who have grown into fine young men.

Jeffrey C. Posnick

Foreword

When I was asked by Jeff Posnick to write a foreword to his new book on orthognathic surgery I said yes, without hesitation. I have known and respected Jeff since he was an enthusiastic, curious, and very bright student at the Harvard School of Dental Medicine, class of 1977. These formative years as a student were instrumental in the subsequent development of his interests and career in craniofacial and orthognathic surgery. Jeff would attend the Children's Hospital Craniofacial Clinic on many a Friday afternoon where he would observe Joseph Murray, John Mulliken, and me interacting with complex craniofacial patients, their families, our students and residents and the rest of our team. From the beginning, I liked Jeff personally, and I developed a relationship with him because he was very attentive and asked many probing questions. He did this not to show off (as Harvard students have been known to do) but to satisfy his inherent curiosity and eagerness to learn. To this day, Jeff will occasionally call to discuss a case and ask for advice, not because he does not have his own ideas or because he does not know what to do, but to check with someone else to see if that person has something to add. This is consistent with his desire to obtain the best information to help each patient and to educate himself. When you give Jeff advice, he always has probing follow-up questions to test your knowledge and recommendations. I have very much enjoyed these interactions over the years, even when our opinions have differed. I also admire Jeff's persistent "sense of wonder" during his long career.*

Never having written a Foreword before, I considered the role of the Foreword and Foreword writer. I was surprised to find that most texts in surgical disciplines have a Preface or an Introduction written by the author, telling how the author became interested in the subject of the book and describing how the book came about. The Preface or Introduction may also contain a summary of the contents of the book. The less common Foreword, on the other hand, is a short introductory statement written by someone other than the author. The writer of a Foreword may be an expert in the field, an author of a similar book and may have a relationship with the author. Presumably, good things will be said about the book, and the author of the Foreword will tell the reader why reading the book is worthwhile. In this respect, the Foreword may be helpful to the publisher for the purpose of marketing.

The more I began thinking about the task at hand, the more onerous it seemed to become. There was no doubt in

my mind that this book would be a major contribution to the field, as was Jeff Posnick's 2-volume book: *Craniofacial and Maxillofacial Surgery in Children and Adolescents*, Philadelphia, WB Saunders, 2000. Dr. Paul Tessier wrote in his Foreword to that book: "Thanks go to Dr. Posnick for his overall work and to the publisher for accepting such an abundance of images for printing. As we approach the year 2000 (which has no quantitative reality), this book is already a landmark in craniofacial surgery." M. Michael Cohen Jr. wrote a second foreword calling it a "tour de force" and noting that Jeff wrote 40 of 45 chapters, making it an unusual single-authored book relative to the primary subject. Well, *Orthognathic Surgery: Principles and Practice* is equally a "landmark" and a "tour de force" and there is no use in trying to say something clever about it. Anyone who reads this book will find that it speaks for itself: "*Res ipsi loquitur.*"

As with *Craniofacial and Maxillofacial Surgery in Children and Adolescents*, *Orthognathic Surgery: Principles and Practice* is a single-authored, 2-volume set and therefore has a consistent format, writing style, and "personality" not usually achieved in a multi-authored and edited textbook. This makes it easier and more pleasant to read. The book is divided into seven sections: Basic Principles and Concepts; Planning, Surgical Technique, and Complications; Classic Patterns and Presentations of Dentofacial Deformities; Frequently Seen Malformations with Dentofacial Deformity; Cleft Jaw Deformities; Post-Traumatic Dentofacial Deformities; and Frequent Aesthetic Considerations in the Dentofacial Deformity Patient. Dr. Posnick wrote 39 of the 40 chapters. The first and only invited chapter is the wonderfully informative and entertaining contribution by Jeff's long-term friend and colleague, M. Michael Cohen Jr.: "*The New Perspectives on the Face.*"

Jeff Posnick is meticulous and pays obsessive attention to detail. Therefore, each chapter includes comprehensive background material presented with a scholarly review of the pertinent literature. The relevance of this background to the overall treatment planning, execution, and outcome of orthognathic surgery is revealed and all this is supported by the incredible, well-documented, and beautifully illustrated material from Jeff's personal experience and practice. This presentation allows the reader to benefit from Jeff's thinking and his triumphs, challenges, and difficulties.

It is not the role of the Foreword writer to summarize the book. However, I would like to describe the highlights of just two chapters to support my laudatory comments above. Chapter 2 is an account of the pioneers in orthodontics, oral and maxillofacial surgery, plastic surgery, and

*Mulliken JB: A Sense of Wonder, *Plast Reconstr Surg* 110:1353-1359, 2002.

craniofacial surgery. Jeff Posnick painstakingly chronicles the critical advances in these specialties that brought us to our current state. The chapter reads like an exciting novel. Not only is the history documented in referenced detail, but also anecdotes of personal relationships between these great leaders and personal communications regarding their thinking, ideas, triumphs, and tribulations are described. The chapter ends by thanking the pioneers for their contributions and an appeal to future generations of surgeons to take up the challenge of creating their own innovations.

Chapter 28 on hemifacial microsomia (HFM), a deformity in which I am particularly interested, is another example of the quality of this text. Jeff Posnick and I have some disagreements in this area, particularly regarding the natural progression of the deformity and timing of treatment. We also disagree about the potential benefits of operative correction during growth, i.e., in the mixed dentition stage. Nevertheless, this chapter is one of the most comprehensive treatises on the condition, what is known of the etiopathogenesis and all the significant issues related to the care of patients with this variable, and in my opinion, progressive facial asymmetry that you will find in one location. He has reviewed the pertinent literature, and presented and critically evaluated the available data. By doing this, he implies the importance of understanding the natural history of the deformity and the patterns of growth in the management of these patients. Jeff Posnick is also correctly cautious and skeptical about the use of distraction osteogenesis for early correction. My experience is somewhat different.

However, Jeff presents the facts as he sees them, and the conclusions are debatable but fair.

Much has been written on the subject of orthognathic surgery from its history, basic biology and physiology of the operations, descriptions of the techniques, peer-reviewed outcome studies, to review articles and textbooks. The challenge in writing about a common subject is to bring new insights and information to the readers; to say something new or significant and not to simply say what has already been said. Jeff Posnick meets this challenge in *Orthognathic Surgery: Principles and Practice*. It is comprehensive, well referenced, data supported, and scholarly. Also of note is the amazing number of quality color illustrations, a credit to Jeff and to the commitment of the publisher.

I started this project on a beautiful, early summer weekend in Boston, thinking I would skim the chapters quickly for a few hours to get a feel for the book. Not by choice or plan, however, I spent the entire weekend reading the book; I could not put it down. I suspect the readers will have the same experience. This text should be required reading for all surgeons interested in orthognathic surgery.

Leonard B. Kaban, DMD, MD

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Preface

The treatment of dentofacial deformities has come a long way since 1897 when Vilray Blair, with Edward Angle's coaxing, completed bilateral body osteotomies under chloroform anesthesia to setback a prognathic mandible and establish an improved occlusion. The 70-minute operation conducted at the Baptist Hospital in St. Louis, Missouri, also included placement of a custom gutta-percha interocclusal splint and application of intermaxillary fixation.

The field of orthognathic surgery advanced by small increments over the next 6 decades until Hugo Obwegeser executed what has now become the three classic orthognathic procedures: Le Fort I (maxillary) osteotomy with down-fracture and disimpaction; intraoral sagittal split ramus osteotomies of the mandible; and the intraoral oblique osteotomy of the chin. His published results in the 1950s and presentations throughout the 1960s disseminated this early work. The animal model research carried out by William Bell confirmed the safety of these osteotomies and set the stage for refinements in orthognathic procedures by practicing surgeons. During this same timeframe, Hans Luhr boldly challenged standard thinking of osteotomy and fracture healing and stabilization techniques with his concepts of rigid metal plate and screw fixation. Simultaneously, Paul Tessier's imaginative introduction of craniofacial surgery energized thinking concerning the reconstruction of all head and neck conditions.

Today, knowledge of how to safely improve the quality of life for the individual with a dentofacial deformity is extensive. The object is no longer limited to achieving short-term improved occlusion. Currently, the triad of improved quality of life by achieving long-term dental health, enhanced facial aesthetics, and an open airway represent standard thinking. There still remain limitations relating to the uneven geographic distribution of experienced dedicated clinicians and the financial barriers to the correction of dentofacial deformities. However, the value of treatment to improve lives is undisputed.

The last comprehensive textbook on the subject—*Surgical Correction of Dentofacial Deformities* edited by Bell, Proffit, and White (1980)—had a major impact on patient care and remains a landmark in the field. Since then, other published texts have been useful but not comprehensive. After setting the outline for this project, my initial intention was to have experts in the field make contributions. I soon realized this was impractical if a consistent and comprehensive level of cohesive knowledge on the subject was to be compiled in a timely manner. In writing this single-authored text (the exception being a chapter contribution by M. Michael Cohen Jr.), I enlisted the help of clinicians from a spectrum of specialties to read each chapter for accuracy, adequacy of depth, and readability. This included critiques from academicians, clinicians in practice, past surgical fellows, and residents in training. They came from a spectrum of specialties, including oral and maxillofacial surgery, orthodontics, periodontics, prosthodontics, speech pathology, otolaryngology/head and neck surgery, plastic surgery, anesthesiology, medical genetics, sleep medicine, radiology, psychology and psychiatry, and pathology. I am grateful for their suggestions, as each brought a different perspective and individual criticism. By clarifying current knowledge on the subject, I hope this text encourages quality care and further advances in the field.

I would also like to thank my patients who have allowed the use of their case studies as teaching instruments. The presentation of clinical problems and real-life solutions remains an invaluable way to convey this knowledge. Their contributions will no doubt minimize treatment errors and optimize results for future patients.

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▶ Orthognathic Surgery Videos

Go to www.ElsevierOrthognathicSurgery.com to view the videos

INTRA-NASAL PROCEDURES

Video 1: Septoplasty and reduction of the inferior turbinates carried out through Le Fort I downfracture

Video 2: Septoplasty and reduction of the inferior turbinates carried out through Le Fort I downfracture

Video 3: Recontouring of the nasal floor, pyriform rims and anterior nasal spine region carried out during Le Fort I osteotomy

Video 4: Recontouring of the nasal floor, pyriform rims and anterior nasal spine region carried out during Le Fort I osteotomy

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Video 6-3: Maxillary Le Fort I osteotomy

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Video 6-5: Removal of impacted maxillary wisdom teeth

Video 6-6: Continuing the Le Fort I osteotomy: removal of bony interferences, placement of the intermediate splint, and securing intermaxillary fixation

Video 6-7: Achieving the desired vertical midface dimension

Video 6-8: Placement of titanium plate and screw fixation across Le Fort I osteotomy

Video 6-9: Recontouring of the nasal floor, pyriform rims and anterior nasal spine region

Video 6-10: Release of IMF—confirming the correct occlusion-removal of the intermediate splint-securing of the final splint

Video 6-11: Osseous genioplasty

Video 6-12: Splitting of each sagittal ramus osteotomy of the mandible and removal of impacted mandibular wisdom teeth

Video 6-13: Securing of intermaxillary fixation through the final splint

Video 6-14: Insertion of the transbuccal trocar, seating of the proximal segment, removal of bony interferences, and placement of bicortical screw fixation

Video 6-15: Release of intermaxillary fixation and confirmation of correct occlusion

Video 6-16: Wound closure

Patient 2

Video 7-1: Patient preparation and draping

Video 7-2: Sagittal split ramus osteotomies: Incision placement, dissection, and cortical cuts

Video 7-3: Maxillary Le Fort I osteotomy

Video 7-4: Septoplasty and reduction of the inferior turbinates

Video 7-5: Continuing the Le Fort I osteotomy: removal of bony interferences, placement of the intermediate splint, and securing intermaxillary fixation

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1

The New Perspectives on the Face⁺

M. MICHAEL COHEN JR.

- What Constitutes a Face?
- Facial Perspectives and Stages in the Life Cycle
- Evolutionary Considerations
- Craniofacial Growth and Development
- Dymorphic Faces
- The Outer Limits in Facial Surgery
- Making Blind People See Again
- Prosopagnosia
- The New Psychiatric Genetics
- Artistic Perspective

What Constitutes a Face?

Structural Definitions

There are several ways to view faces. Figure 1-1 shows two fish.¹⁴ If two eyes and a mouth define a face, then fish have faces. They may have one or two nostrils, which are not connected to the mouth. However, water entering the nostrils does bathe the olfactory mucosa. Fish lack an ear canal, but the inner ear is present. Weber's bones connect the swim bladder to the inner ear, and sound is transmitted.¹⁰

Another definition requires evolutionary transformation of the skull, in which a face is recognizable in mammals, but not in fish, amphibians, or reptiles (Fig. 1-2). The jaw is suspended from the braincase in reptiles. Mammals, however, have three ear ossicles, a secondary palate separating the airway passage from the mouth, and vertical positioning of the dentary.³⁵

Behavioral Definitions

All animals communicate with each other in various ways by tactile, chemical, visual, and acoustical signaling. Insects have heads, but do they have faces? Paper wasps (*Polistes*

⁺This chapter is dedicated to my Fellow Angela M. Castro, MD.



• Figure 1-1 Two fish faces. Copyright © Shutterstock.com.

dominulus) signal their status to each other by the number of black splotches on their yellow faces (Fig. 1-3).⁴¹

Some define faces by the presence and use of facial muscles, which do not exist in fish, amphibians, reptiles, or birds. In contrast, mammals can suckle and later chew, supported by a muscular tongue and movable lips and cheeks (Fig. 1-4). Often, the muscles in the ears can change positions to aid in hearing; a movable nose for smelling and touching, and facial hair, the *vibrissae*, are associated with musculature and serve as tactile organs.¹⁰

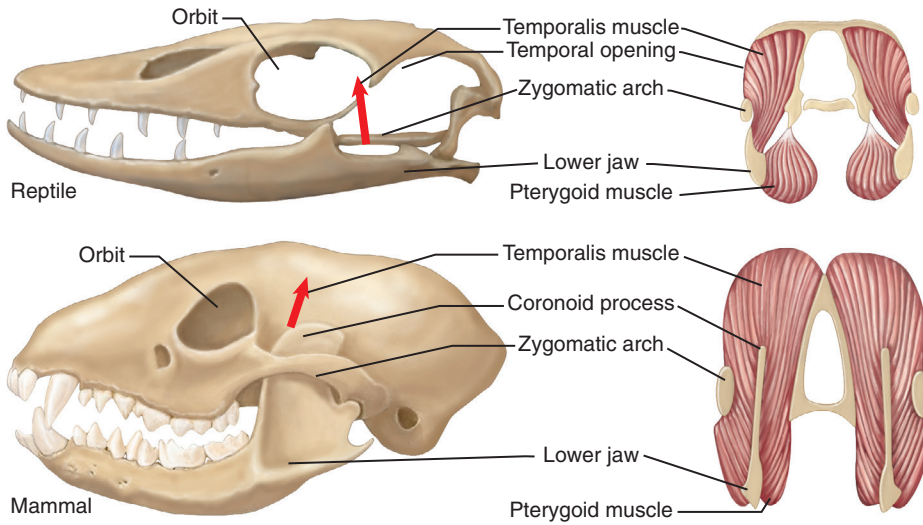
For some, a behavioral definition must include facial expressions, which are found in various primates (Fig. 1-5), including humans (Fig. 1-6).¹⁰

Symbolic Definitions

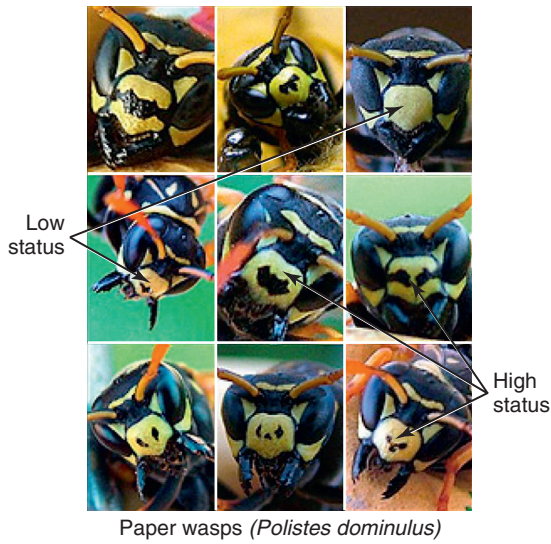
The term *symbolism* has been defined in many different ways. Here, I define *symbolic transformation* as the ambiguous representation of two things by association or resemblance. Two examples are shown in Figures 1-7 and 1-8.¹⁰

Facial Perspectives and Stages in the Life Cycle

Box 1-1 summarizes all possible perspectives from which the face can be described; Box 1-2 lists all the stages in the



• **Figure 1-2** Transformation of a mammalian-like reptile to a mammalian skull. Lateral and cross-sectional views of the skulls. *Based on Radinsky, 1987.*



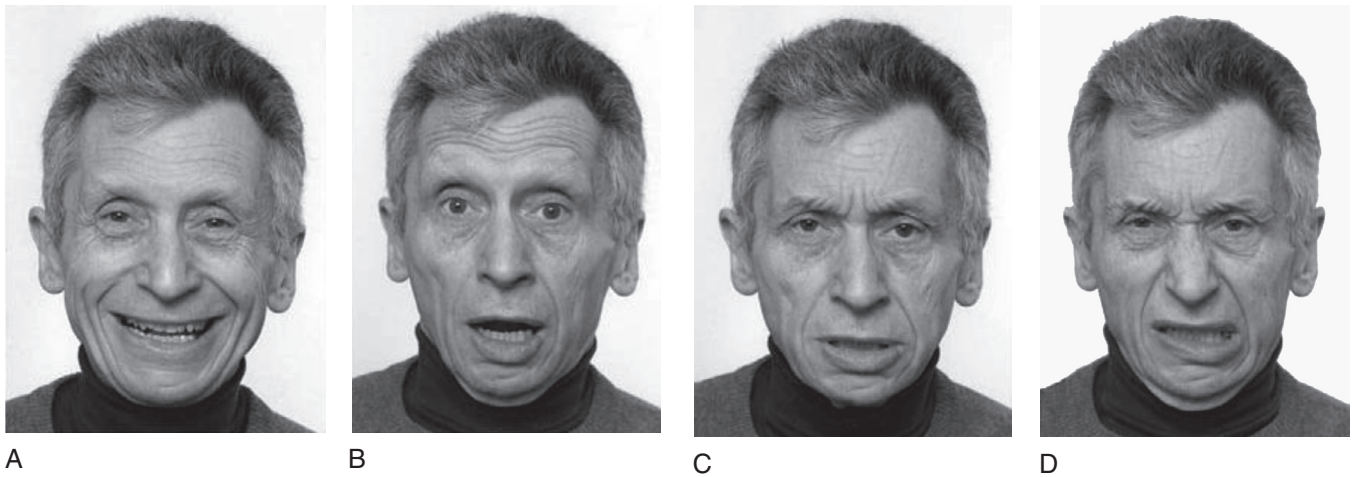
• **Figure 1-3** Wasp faces signal their status to each other by the number of black splotches on their yellow faces. The more blotches, the higher the status. *Copyright 2004 Elizabeth Tibbetts. Modified by M. Michael Cohen Jr.*



• **Figure 1-4** Mammals can suckle and later chew, supported by a muscular tongue and movable lips and cheeks.



• **Figure 1-5** Rhesus monkey facial expressions. **From left to right**, a bared-teeth display, a scream, an open-mouth threat, and a relaxed, open-mouth face (play face). *From Parr and Heintz, 2009.*



• **Figure 1-6** Human facial expressions. **A**, Happiness. **B**, Surprise. **C**, Anger. **D**, Disgust. *Based on Cohen, 2006.*



• **Figure 1-7** Gossiping scallions. *From Elffers, 1997.*



• **Figure 1-8** *All Is Vanity*. The mirror is also a skull. House of Art, New York.

• BOX 1-1

Perspectives on the Face

- Evolutionary
- Developmental
- Anatomic
- Genetic
- Surgical
- Psychological
- Sociocultural
- Artistic

• BOX 1-2

The Face in the Life Cycle

- Intrauterine development
- Birth
- Infancy
- Childhood
- Adolescence
- Adulthood
- Old age
- Death

life cycle; and [Box 1-3](#) lists the origin of some craniofacial components. [Figure 1-9](#) shows embryonic facial development at approximately 42 to 44 days.²⁵ [Figure 1-10](#) shows skull molding in a newborn. [Figure 1-11](#) shows the face of a small child, and [Figure 1-12](#) shows a painting, *The Stages of Human Life*, by Hans Baldung Grien. In this allegory, a young woman, an old woman, and a dead woman are linked by their hands and arms.⁴ The dead woman holds an hourglass timer, indicating that life is over.

• BOX 1-3

Origins of Some Craniofacial Components

Neural Crest Origin

Facial connective tissue
 Facial bones
 Nasal capsule
 Frontal bone
 Metopic suture
 Sagittal suture
 Temporal squama

Mesodermal Origin

Facial muscles
 Endothelial lining of blood vessels
 Chondrocranium
 Parietal bones
 Coronal suture
 Lambdoid suture
 Temporal bones (except squama)
 Occipital bone (somites plus squama)



• **Figure 1-9** Embryo of about 42 to 44 days. The developing face has a frontal area with bulging cerebral hemispheres; a nasodorsal center with nasoseptal and nasozygomatic portions; nasal pits delineated by nasal ridges with premaxillary, medianasal, and latero-nasal portions; maxillary primordia; interpremaxillary depression; a premaxillary–maxillary junction; and a lower jaw. *Courtesy of Jan E. Jirásek, Prague.*

Evolutionary Considerations

Evolution of the Mammalian Brain

The evolution of the mammalian brain is based on the development of the neocortex (Fig. 1-13), which also resulted in a cranial shape that was different than that of the dinosaurs. The coexistence of dinosaurs and mammals and their competition during the Mesozoic era made mammalian neocortical development possible.⁵

Dinosaurs were large and had long life spans of about 100 years. In contrast, mammals were very small and had short life spans, which allowed them to evolve into many



• **Figure 1-10** Molding of the head caused by compression during passage through the birth canal. *From Cohen, 2006.*

different species rapidly. Possibly, there were 50 generations of mammals during one dinosaur generation.

Mammals were nocturnal and hunted at night. With the development of the neocortex, mammals had an active lifestyle as well as acute hearing and smelling, which made insect prey easier to come by and made escape from predators easier as well. The end of the dinosaur era allowed mammals to evolve larger body forms.

Molecular Components in Primate Brain and Craniofacial Evolution

The gene *Microcephalin* (*MCPH1*) was first identified in its mutant form in which it causes primary microcephaly, but the normal gene was then adaptively found to be important in regulating brain size, and it continues to evolve in humans.¹⁸

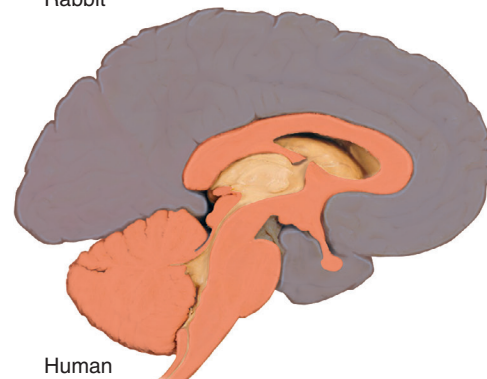
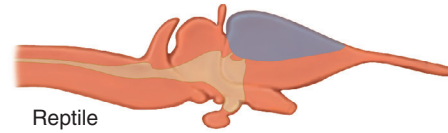
Homozygous *ASPM* mutations also cause microcephaly. The normal gene may regulate neural stem cell proliferation



• **Figure 1-11** Baby's face with relatively large head, prominent eyes, and a diminutive face. From Cohen, 2006.



• **Figure 1-12** *The Stages of Human Life* (1530-1545), by Hans Baldung Grien, oil on wood, 61 × 151 cm, Prado Museum, Madrid. From Buendia, 1989. See text for details. Museo del Prado, Madrid, Spain. Photo credit: Bridgeman-Giraudon/Art Resource, NY.



• **Figure 1-13** Stages in the evolution of the brain. A comparison of the brain size of the fish, reptile, rabbit, and human. As the cerebral cortex increases in size, the cranium becomes larger and more rounded. Based on Campbell, 1970.

and/or differentiation, possibly by mediating spindle-cell assembly during cell division.³¹

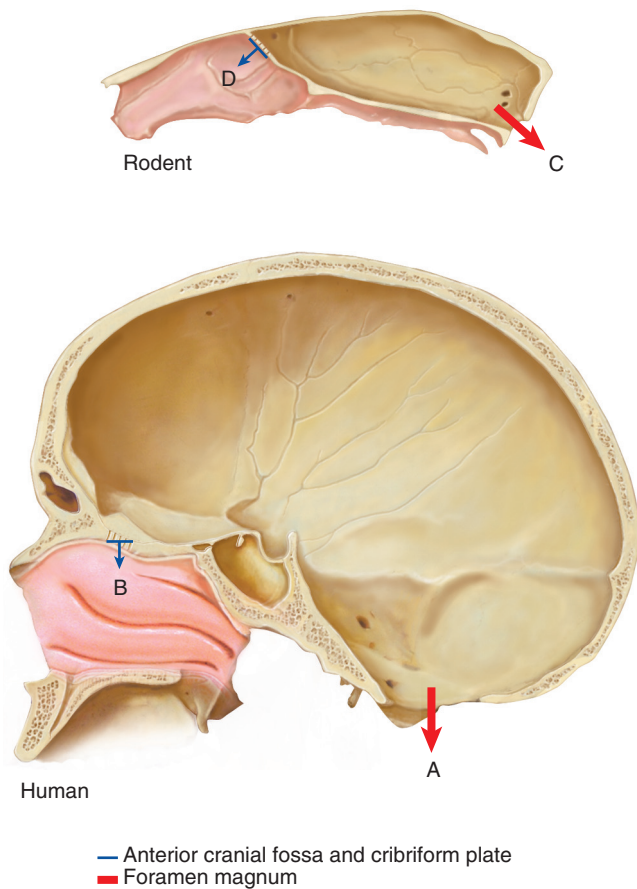
SIGLEC11, a gene involved in sialic acid biology, is expressed in high concentration in microglial cells in the human brain, but only occasionally in the cells of chimpanzees.²⁴

The *Ret finger protein-like 1,2,3 (RFPL 1,2,3)* genes on chromosome 22 are evolutionary forces that play a role in neocortical development.³

GTF2IRD1—a gene that, when mutated, causes craniofacial anomalies—has been shown in its normal form to be a regulatory determinant of craniofacial development.³⁹

Comparison of Different Skulls

Figure 1-14 shows the orientation of the foramen magnum and the anterior cranial base in a rodent and a human. The foramen magnum is posteriorly placed in the rodent and vertically placed in the human. The anterior cranial floor and the cribriform plate face forward in the rodent and are vertically placed in the human.¹⁶ **Figure 1-15** shows a dog's



• **Figure 1-14** Orientation of the foramen magnum and the anterior cranial base in a human and a rodent. The foramen magnum is vertically placed in the human (A) and posteriorly placed in the rodent (C). The anterior cranial floor and the cribriform plate face downward in the human (B) and forward in the rodent (D). Based on Enlow, 1968.

skull compared to a Chevrolet Corvette in contrast to a human skull compared to a camper, indicating an expanded, upright forehead above the face.¹⁷

In [Figure 1-16](#), expansion of the brain and a reduction in human facial prominence results in a flat facial profile compared with a smaller brain and a more protruding gorilla face, resulting in a sloping facial angle.¹⁰ [Figure 1-17](#) compares the growth pattern of a fetal chimp with that of a human fetus. Both fetuses look similar. However, compared with the adult chimp, the adult human more closely resembles its own fetal pattern. Neoteny—a slowdown in the growth rate with a delay in maturation—has occurred in human evolution.²³

Comparison of *Homo sapiens*, *Homo neanderthalensis*, and *Homo floresiensis*

Evidence now shows that there are three separate species of hominins. [Figure 1-18](#) compares the skulls of *Homo neanderthalensis* with *Homo sapiens*. The braincase of modern humans is relatively shorter, and the forehead is rounder, higher, and has a nearly vertical slope. Neanderthals have large brow ridges, projecting midface, elongated skull,

occipital protuberance, a skull capacity 10% greater than that of modern humans, and a distinctive bony labyrinth not found in humans.²⁷ DNA sequence comparisons show that Neanderthals fall outside the variation of modern humans. Molecular divergence provides a date of over 500,000 years.²⁸

[Figure 1-19](#) contrasts the phenotypes of the different species of hominins. *Homo neanderthalensis* has an elongated skull with a larger endocranial capacity than in humans; a distinctive bony labyrinth not found in humans (not shown); a robust skeleton with a barrel-shaped ribcage; a long superior pubic ramus; long clavicles; thick, bowed femoral shafts; large patellas; and large, round, terminal phalanges (fingers). *Homo floresiensis* has a tiny skull with a small endocranial capacity (380 cm³ to 430 cm³), a chinless mandible, a diminutive body (~1 m in height), long arms in relation to the legs (arms hang almost to the knees), unusual shoulders, a wider pelvis than in humans, and large feet (more than 7½ inches long and out of proportion with the short lower limbs, flat feet, and a stubby great toe).¹⁰

The time spans of the three different species of hominins are (1) *Homo sapiens* from 500,000 years ago to the present time; (2) *Homo neanderthalensis* from 500,000 years ago until 30,000 years ago; and (3) *Homo floresiensis* from 95,000 years ago until 12,000 years ago. Thus, from 95,000 to 30,000 years ago, three different species of hominins occupied the earth at the same time.

Craniofacial Growth and Development

Comparative Skull Size with Age

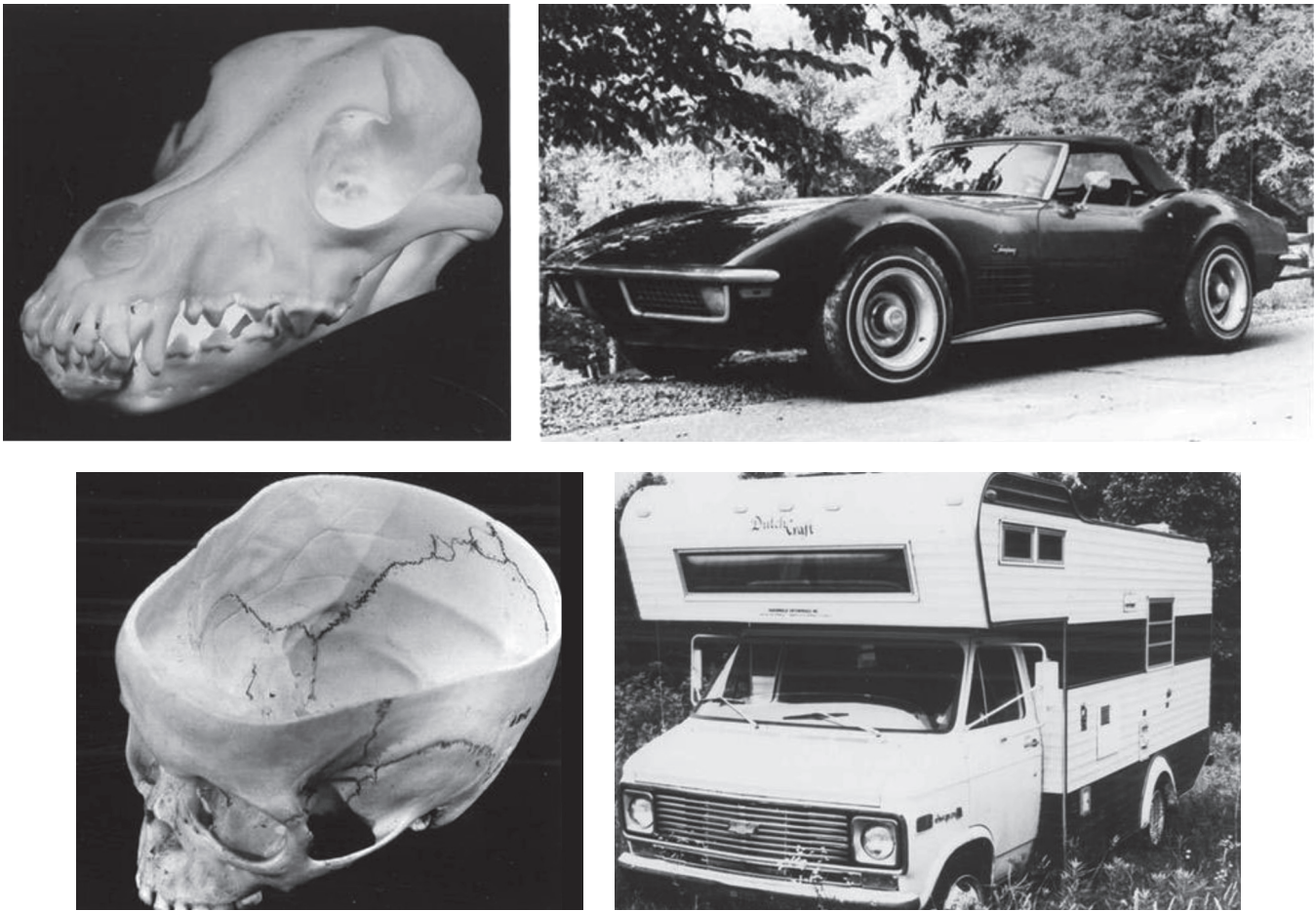
In the newborn skull ([Fig. 1-20](#)), the cranium and the orbits are relatively large, and the face is diminutive. In early childhood, the cranium and the orbits remain relatively large, but the eruption of the primary dentition enlarges the facial skeleton. In the adult skull, the facial skeleton is well-developed with relatively less prominence of the orbits and cranium. The ratio of the cranial volume to the facial volume changes during growth, and these ratios appear in [Figure 1-20](#) under the skulls.¹⁰

Bone Modification with Age

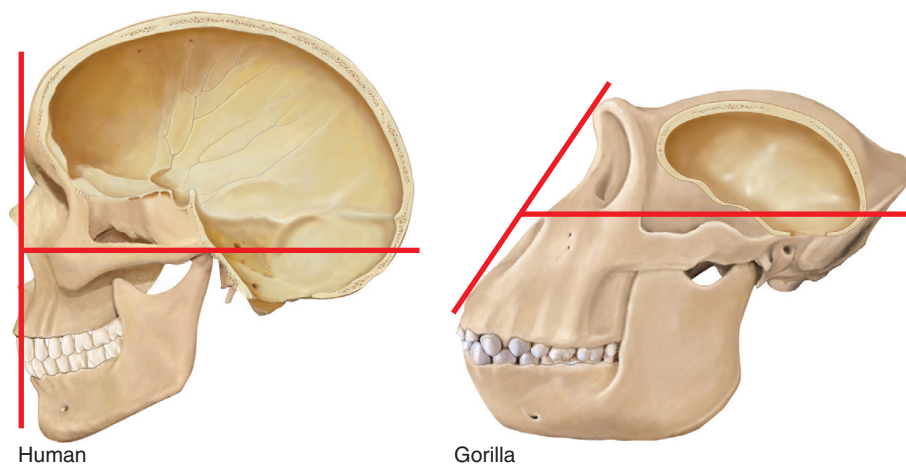
Bone modification of the craniofacial skeleton occurs normally with age (summarized in [Table 1-1](#)). [Table 1-2](#) lists some examples of abnormal alterations in the craniofacial skeleton with age.¹⁰

Alterations in the Face with Age

The changing proportions of head size to body size are illustrated in [Figure 1-21](#). From the 2-month-old embryo to the 22-year-old adult, the relative head size decreases significantly, but it is greatest in the fetus and the infant. [Figure 1-22](#) illustrates line drawings of the facial profile showing alterations from 5 months of age to adulthood, with striking changes in the nose and chin with age.¹⁰ The



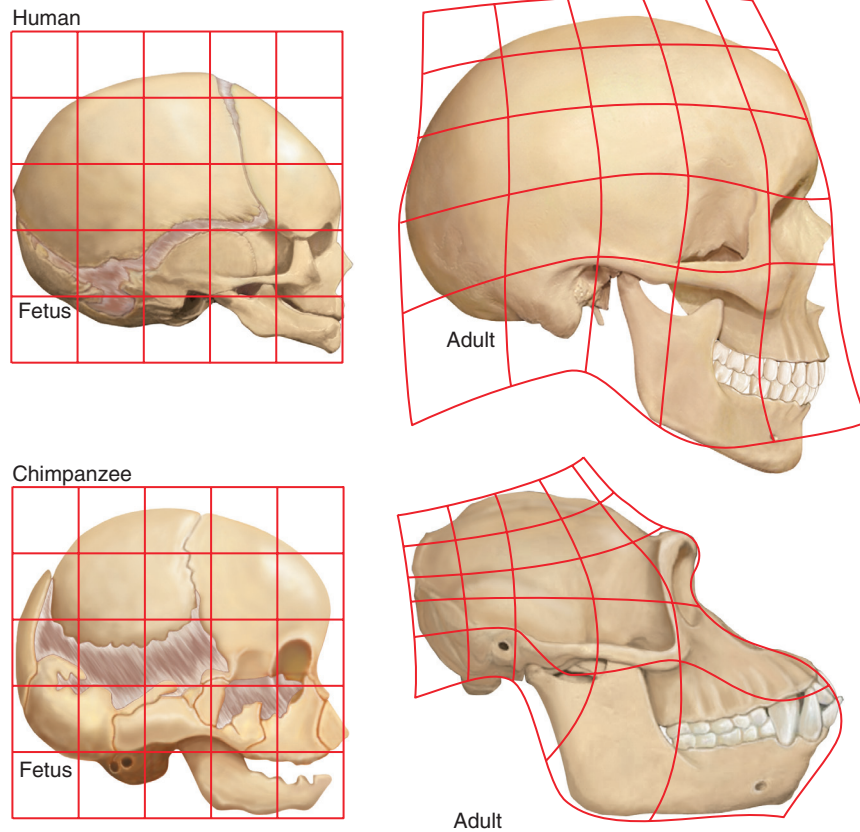
• **Figure 1-15** **Top**, Lateral view of a dog skull compared to a Chevrolet Corvette; the face is anterior to the cranium. **Bottom**, Lateral view of a human skull compared to a camper; the expanded forebrain results in an upright forehead above the face. *From Enlow, 1990.*



• **Figure 1-16** Expansion of the brain and a reduction in the facial prominence of humans result in a flat facial angle compared with the smaller brain and more protrusive face of the gorilla, resulting in a sloping facial angle.

TABLE 1-1 Normal Growth and Modification of the Craniofacial Skeleton with Age

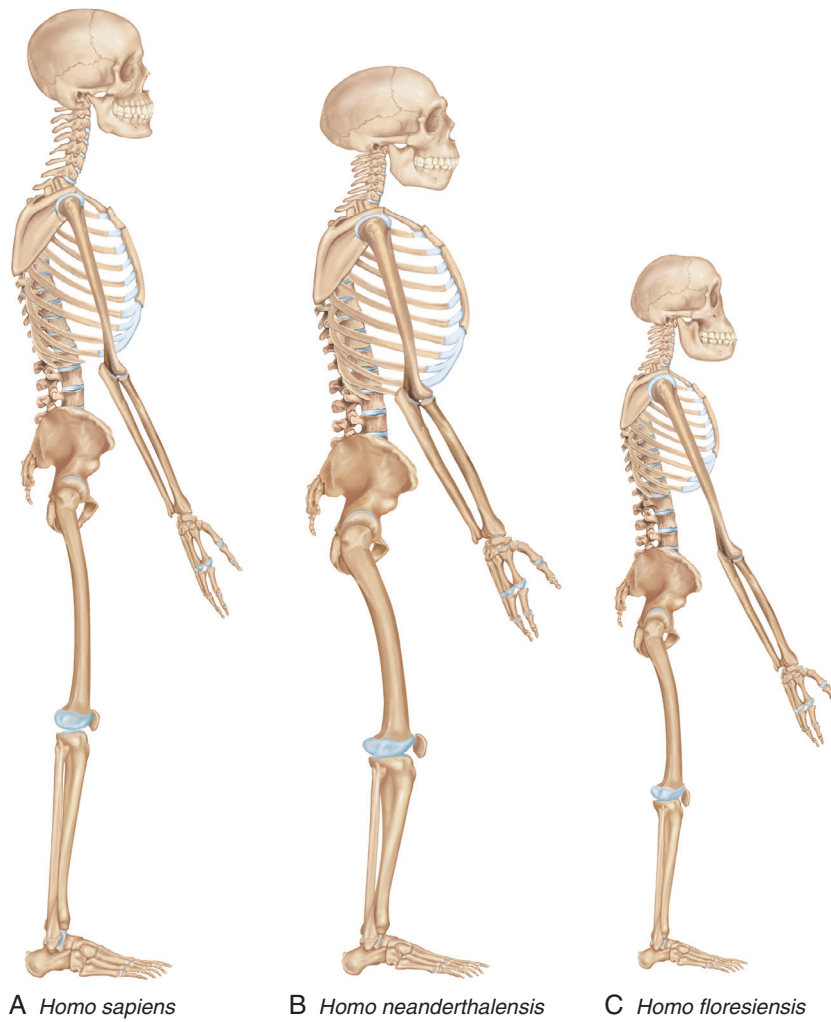
	Cartilaginous Growth	Sutural Growth	Skeletal Mass	Bone Remodeling
Infancy and Childhood	Active	Passive	Increases	Deposition greater than resorption
Adulthood	Absent	Absent	Remains the same	Deposition equals resorption
Old Age	Absent	Absent	Decreases	Resorption greater than deposition



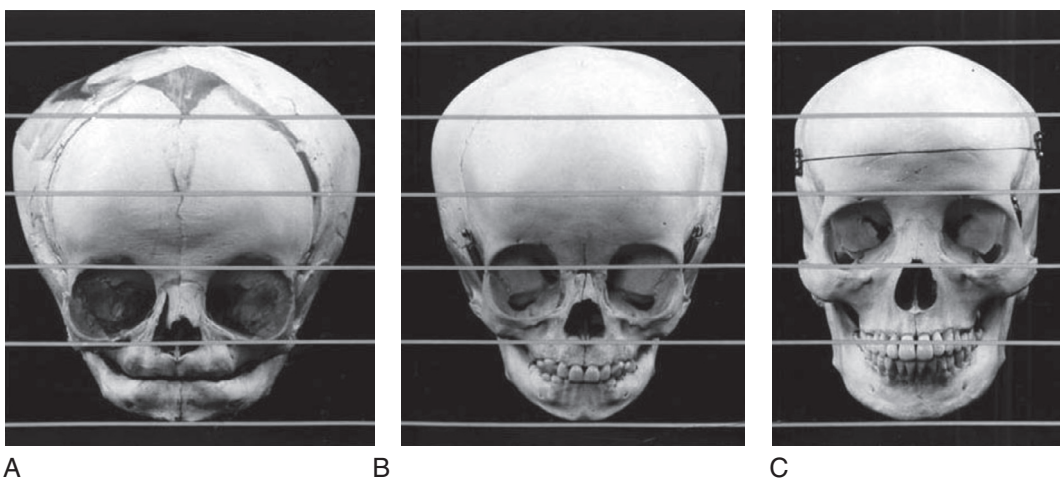
• **Figure 1-17** Morphology of the chimpanzee and human skulls. Compared with the chimp, the adult human skull resembles its fetal counterpart. Neoteny—a slowdown in the growth rate with a delay in maturation—has occurred in human evolution. *Based on Gould, 1977.*



• **Figure 1-18** Lateral view of the skulls of *Homo sapiens* and *Homo neanderthalensis*. See text for description.



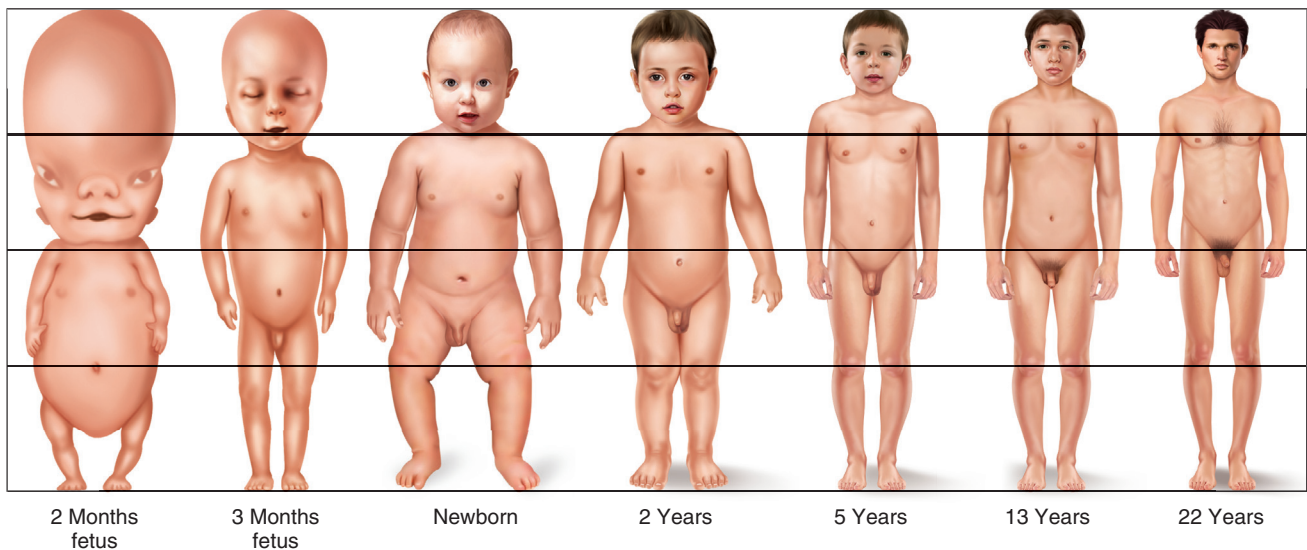
• **Figure 1-19** Comparison of **A**, *Homo sapiens*, **B**, *Homo neanderthalensis*, and **C**, *Homo floresiensis*. See text for description. Based on the work of M. Michael Cohen Jr.



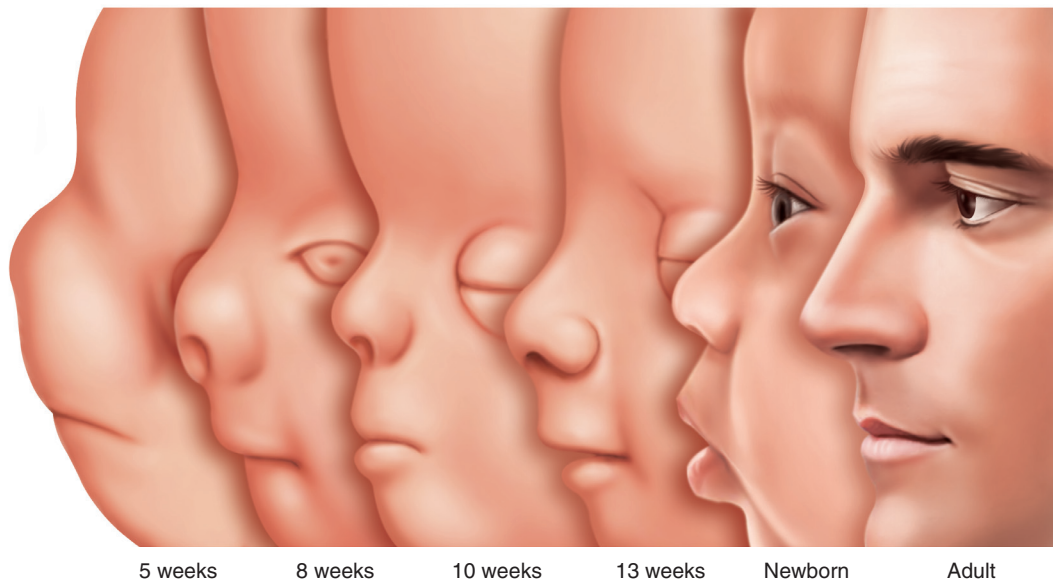
Ratio of cranial volume to facial volume

- Birth (8:1)
- 2 years (5:1)
- 6 years (3:1)
- 12 years (2.5:1)
- 18 years (2:1)

• **Figure 1-20** Alterations of the skull in the newborn, in childhood, and in adulthood. The ratios of the cranial volume to the facial volume change during growth, and these ratios appear under the figure. For a description of skull alterations with age, see text. From Cohen, 2006.



• **Figure 1-21** Diagram illustrating the changing proportions of head size and body size. From the 2-month-old embryo to the 22-year-old adult, the relative head size decreases significantly, but it is greatest in the fetus and the infant. *Based on Scammon, 1953.*



• **Figure 1-22** Line drawing of the human profile illustrating alterations in form and proportion from 5 weeks prenatally through the newborn period to the adult. *Based on Scammon, 1953.*

lower jaw in the male Atlantic salmon is extraordinary. There is rapid and pronounced growth *during adult life* (Fig. 1-23), when the *starving* salmon migrate upriver for spawning.⁴²

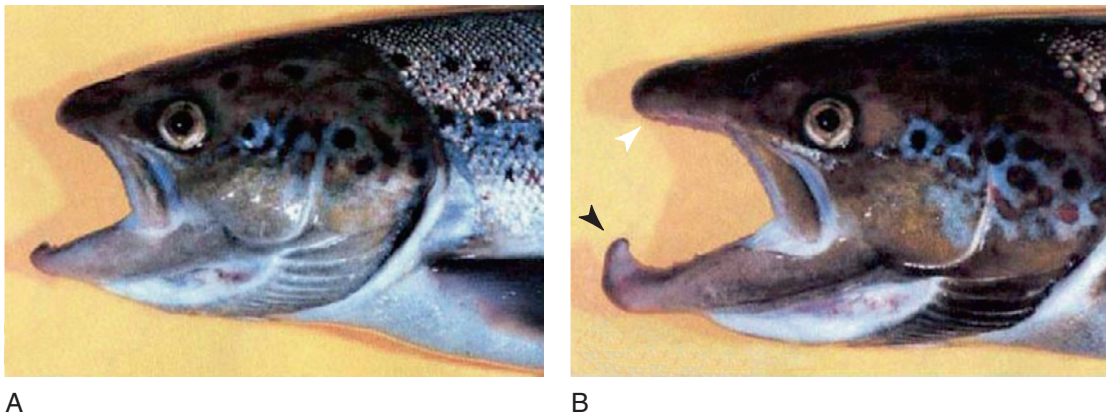
Facial Asymmetry

Those psychologists who study faces and state that “beautiful faces are symmetric” don’t know what they’re talking about, because all normal faces are asymmetric. With respect to the normal face, subtle degrees of asymmetry become particularly evident when properly oriented frontal

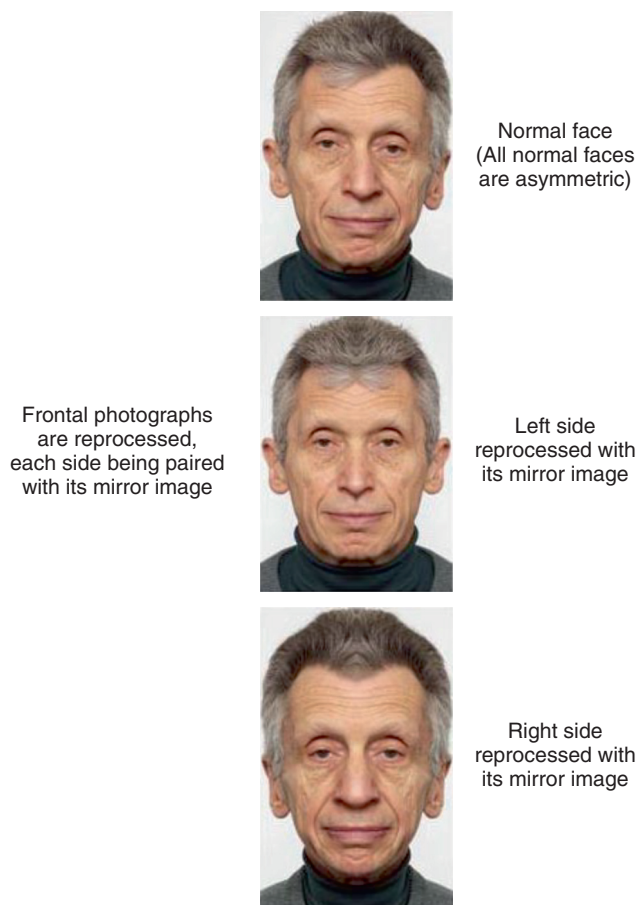
photographs are divided along the median plane and reprocessed, each side being paired with its mirror image (Fig. 1-24).¹⁰

The Skull as a Community of Bones versus the Skull as a Community of Joints

According to Pruzansky,³⁴ the skull is a community of bones separated by joints, but according to Moffett³² the skull is a community of joints separated by bones. Which view is correct? They are simply different contexts in which to view the development of the skull.¹⁰



• **Figure 1-23** Rapid and pronounced growth of the lower jaw in a starving adult male salmon migrating upriver for spawning. **A**, Normal adult male jaw. **B**, Extraordinary upriver growth of the lower jaw. From Witten et al, 2003.



Frontal photographs are reprocessed, each side being paired with its mirror image

Normal face
(All normal faces are asymmetric)

Left side reprocessed with its mirror image

Right side reprocessed with its mirror image

• **Figure 1-24** Demonstration of the facial asymmetry that is so typical of everyone. From Cohen, 2006.

Pruzansky³⁴ stated the following:

“The skull is a community of bones and organ systems of diverse phylogenetic origin and variable patterns of development, altogether relating to several functions vital to the life and well-being of the organism. If in the course of development one member of this community is affected adversely, inevitably other parts will suffer.”



• **Figure 1-25** Midface deficiency secondary to hypoplasia of the nasal capsule in achondroplasia. From Cohen, 2000.

Achondroplasia, caused by several *FGFR3* gain-of-function mutations (particularly Gly380 Arg⁹) illustrates what Pruzansky³⁴ meant (Fig. 1-25): if one member of the community is affected, inevitably other parts will suffer. In this case, the nasal capsule and cranial base, both cartilaginous in origin, have a secondary effect on membrane bones, because all craniofacial bones articulate with one another. Thus, in achondroplasia, the hypoplastic nasal capsule and short anterior cranial fossa result in midface deficiency.^{7,9,10}

The skull may also be considered a community of joints separated by bones. Types of joints, known as *craniofacial articulations*, include synovial, cartilaginous, fibrous, and dental.³² Table 1-3 summarizes craniofacial articulations together with their physiologic and mechanical functions and their remodeling responses.¹⁰

TABLE 1-2 Some Examples of Abnormal Alterations in the Craniofacial Skeleton with Age

	Abnormal Skeletal Condition	Pathologic Process
Infancy and Childhood	Achondroplasia	Bony midface deficiency secondary to hypoplasia of cartilaginous nasal capsule and cranial base
	Craniosynostosis	Premature sutural fusion by bone deposition
Adulthood	Acromegaly	Bone deposition dramatically exceeds bone resorption
	Hemifacial atrophy	Localized bone resorption exceeds bone deposition
Old Age	Paget disease of bone	Bone deposition exceeds bone resorption
	Senile osteoporosis	Bone resorption exceeds bone deposition

Dysmorphic Faces

Four different types of faces are contrasted in [Figure 1-26](#): (1) the normal face; (2) Down syndrome with minor anomalies; (3) Crouzon syndrome; and (4) a bizarre prenatally determined face. Three views of Williams syndrome are illustrated in [Figure 1-27](#). The combination of minor facial anomalies—strabismus, anteverted nares, and thick lips—are evident in all three patients. In contrast, the severe facial anomalies associated with holoprosencephaly—cyclopia, ethmocephaly, cebocephaly, and premaxillary agenesis—are shown in [Figure 1-28](#).^{6,8,10-12} A newborn with diprosopus—a double face with two nostrils, four eyes, two noses, and two mouths—is illustrated in [Figure 1-29](#). The most bizarre case of all is shown in [Figure 1-30](#). This patient is missing his lateral orbital walls, and he is able to pull his eyes apart laterally without ripping his optic heads from the back of his eyeballs. How he does this is mysterious, but I suspect he started doing this very early in childhood and gradually increased the distance over time.

The Outer Limits in Facial Surgery

The problems surrounding complete facial transplantation are technical, psychological, and ethical. Earlier, facial transplantation was rejected by many institutions, with the exception of the Cleveland Clinic's Institutional Review Board, and I have discussed the concept of facial transplantation and its associated problems elsewhere.¹⁰ Although it was rejected earlier by the French National Ethics Advisory Committee, the door was left open for a severely disfigured woman bitten by a dog for which she received a lower facial transplant. Since then, at least seven facial transplants have been carried out. The result of a facial transplant after a gunshot wound is shown in [Figure 1-31](#).

Making Blind People See Again

The cornea covers the eye globe and a narrow zone between the cornea and the conjunctiva known as the *limbus* ([Fig. 1-32](#)), which is a source of stem cells for corneal epithelium.

In recent years, new techniques that can restore vision in certain types of blindness have come to the fore. In a surgical procedure called *limbal cell transplantation*, extraction of stem cells from the healthy contralateral eye of a patient or from a relative in the family is transplanted into a patient's eye with corneal degeneration, blindness, or some other ocular disease. The stem cells then differentiate into corneal epithelial cells and improve vision.¹⁹ The limbus itself can be destroyed by chemical burns, by thermal burns, or by infection, which results in corneal stem-cell deficiency. However, it has been recently reported that a viable alternative source of cells for transplantation consists of limbal cells maintained in culture.³⁶ A biosynthetic cornea created from human collagen has also been developed. It mimics the protein's scaffolding and can be used to trigger regeneration of a patient's own corneal cells.²⁰

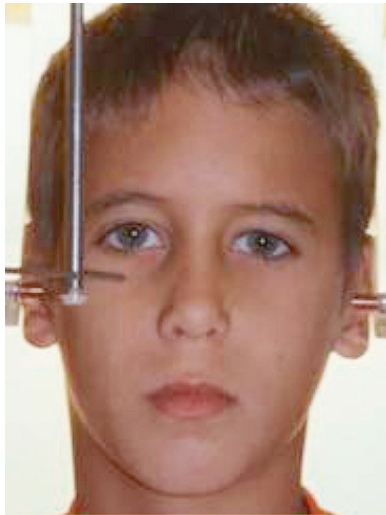
Age-related macular degeneration is a major cause of blindness. Although there is no treatment for the avascular type, the neovascular type results from an imbalance in antiangiogenic and proangiogenic factors and can be treated. Intravitreal injection of VEGF inhibitors results in significant recovery of vision in 30% to 40% of patients.⁴⁰

Leber congenital amaurosis is an inherited group of rod-cone dystrophies resulting in congenital blindness. One form caused by the *RPE65* gene accounts for 16% of the cases. Using rAAV as a vector for retinal gene therapy, a cannula is passed through the front of the eye and across the vitreous gel. Working copies of the gene are injected into the back of the eye. Maguire and colleagues³⁰ reported improvement in visual acuity for all patients.

Several animal studies show great promise for future treatment. The development of retinal cell transplants has been studied in rabbits,³⁸ and retinal repair by transplantation of photoreceptor precursors has been studied in mice.²⁹

Prosopagnosia

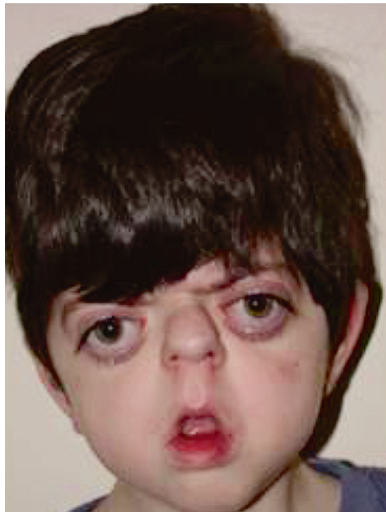
The ability to identify human faces is remarkable. Although the face ages with time, it is possible to instantly identify a face of someone who has not been seen for 25 or 30 years. An area in the brain that governs facial recognition is found



Normal face



Down syndrome



Crouzon syndrome



Severe prenatal anomaly

- **Figure 1-26** Four kinds of faces with different implications. A normal child's face is shown. Down syndrome involves many minor facial anomalies that are diagnostic. Crouzon syndrome is severe but can be treated successfully with surgery. There is no effective treatment for the infant with severe facial anomalies and hydrocephalus. *Based on the work of M. Michael Cohen Jr. Crouzon syndrome is by courtesy of Bonnie Padwa, Boston.*



- **Figure 1-27** Three patients with Williams syndrome of different ages. All have strabismus, anteverted nares, and thick lips. *From Cohen, 1997, 2002, 2006, and 2007, a and b.*

Holoprosencephaly



Cyclopia



Ethmocephaly



Cebocephaly



Premaxillary agenesis

• **Figure 1-28** Faces associated with holoprosencephaly. From Cohen, 1997, 2002, 2006, and 2007, a and b.



• **Figure 1-29** Diprosopus. Courtesy of Gosla Reddy.



• **Figure 1-30** Hypertelorism with absent lateral orbital walls. This man is able to manipulate his eyes without ripping his optic heads from the back of his eyeballs. I suspect that he learned to do this very gradually. Courtesy of M-Reza Farahvash.



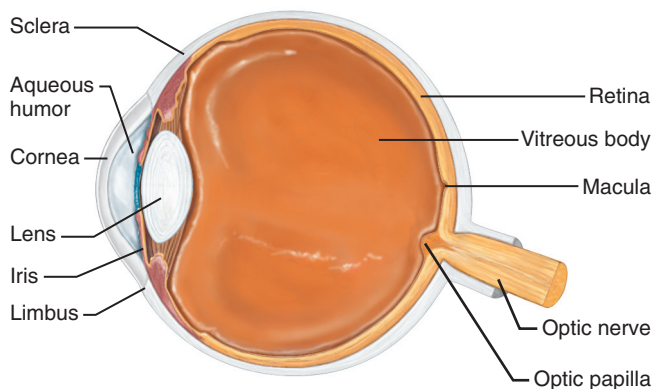
• **Figure 1-31 A**, Three frontal views by dates. 12/3/08: A 45-year-old woman with a shotgun blast to the face. She underwent a nearly total facial transplantation, including a composite LeFort III midfacial skeleton, overlying skin, soft tissue, nose, lower eyelids, upper lip, total infra-orbital floor, both zygomas, both parotid glands, the anterior maxilla with central maxillary incisors, the whole alveolus, the anterior hard palate, and intraoral mucosa. Note tracheostomy. 5/4/09: Second facial transplantation with construction of the nose and lips. 8/23/10: Reduction of facial fat and better facial appearance. **B**, Three profile views with the same dates as in **A**. 5/4/09: Note better projection of the nose. 8/23/10: Mandibular advancement with normal lower facial appearance. Both the upper and lower portions of the face are balanced. Courtesy of the Cleveland Clinic.

12/3/08

5/4/09

8/23/10

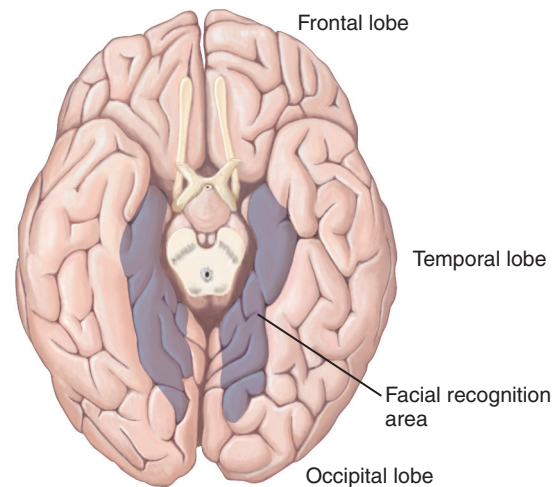
at the base of the cerebral cortex (Fig. 1-33). Any lesion that destroys this area impairs the ability to identify faces but has almost no other effects, although the dimming of vision may occur in some affected individuals. The patient can read, name objects, and match a full-face picture with a profile picture of the same person. Only the ability to recognize specific faces is lost; this is a neurological condition known as *prosopagnosia*. An affected man who cannot recognize his own wife can see her, but he can only recognize who she is when he hears her speak.²¹ An epidemiologic study has found a prevalence rate of prosopagnosia of 2.47%; of 17 cases, 14 subjects had at least one first-degree relative with prosopagnosia.²⁶



• **Figure 1-32** Eye globe, horizontal plane. Note the limbus. Based on Cohen, 2006.

The New Psychiatric Genetics

The brain, as part of the craniofacial complex, is associated with a number of psychiatric and neurodegenerative disorders. In medical genetics, we used to delineate newly recognized syndromes by describing them as single entities and then finding a Mendelian pattern, a chromosomal anomaly, or a teratogenic cause. After molecular genetics became established, it became clear that the same disorder might



• **Figure 1-33** Prosopagnosia. Note the area of the brain that governs facial recognition found at the base of the temporal and occipital lobes on both sides of the cerebral cortex. Based on Geschwind, 1979.

TABLE 1-3 Craniofacial Articulations

Type of Articulation	Example	Physiological Function	Mechanical Function	Remodeling Response
Synovial	Temporomandibular joint	Jaw movement	Resists compression and shear to some extent	Limited, avascular
Cartilaginous	Cranial base synchondroses	Active growth	Resists compression	Limited, avascular
Fibrous	Cranial sutures	Allows passage through narrow birth canal; growth secondary to brain enlargement	Respond to tension	Great, vascularized
	Facial sutures	Mastication	Sutures remain patent until old age; shock absorbers for forces of mastication	Great, vascularized
	Periodontal fibers	Eruption of teeth; anchoring support of teeth	Responds to tension, compression, and shear	Great, vascularized
Dental	Occlusal and interproximal articulations	Mastication and speech	Subject to compression and shear	None, acellular

have causative mutations in several different genes on different chromosomes. For example, Biedl-Bardet syndrome can be caused by mutations in 14 different genes, all on different chromosomes. Thus, we learned that what appeared to be a single disorder was etiologically heterogeneous.

Because psychiatric thinking began by attributing behavioral problems to earlier psychological events, it was the last bastion to be stormed by molecular genetics. Behavioral disorders are now being delineated at a molecular level, but, like the early delineation of physical genetic disorders, progress in psychiatric genetics only accounts for a small percentage of cases at present. Disorders with molecular findings include intellectual disability, schizophrenia, bipolar disorder, Alzheimer disease, autism, Parkinson disease, Huntington disease, and frontotemporal dementia.¹³

Artistic Perspective

Figure 1-34 compares two Renaissance paintings: a Florentine *Portrait of a Man with a Medal of Cosimo the Elder* by Botticelli and a Venetian *Portrait of Jacopo Soranzo* by Tintoretto. The difference in painting style is striking. In the Florentine painting, the outlines of the face are sharply drawn with a three-dimensional sculptural quality together with a sense of palpability. In contrast, in the Venetian painting, the lines of the face are soft and less distinct but with a sense of atmosphere surrounding the figure.²



• **Figure 1-34** **Left**, Close-up of a Florentine *Portrait of a Man with a Medal of Cosimo the Elder* by Botticelli. **Right**, Close-up of a Venetian *Portrait of Jacopo Soranzo*, by Tintoretto. For stylistic interpretation, see text. **A**, From the Uffizi Gallery, Florence Inv 1890 no. 1,488; **B**, from Gallerie del' Accademia, Venice. Image source: Art Resource, NY.

References

- Arneson DJ: *Stories: Tweety and Sylvester*, Middleton, Conn, 1971, Xerox Education Publications. Courtesy of Warner Bros.
- Berenson B: *Italian Painters of the Renaissance*, New York, 1976, Meridian.
- Bonnefont J, Nikolaev SI, Perrier AL, Guo S, Cartier L, Sorce S, Laforge T, Aubry L, Khaitovich P, Peschanski M, Stylianou E, Antonarakis E, Karl-Heinz K: Evolutionary forces shape the human *RFPL 1,2,3* genes toward a role in neocortex development. *Am J Hum Genet* 83:208–218, 2008.
- Buendia JR: *Basic Guide to the Prado*, Madrid, 1989, Silex.
- Campbell B: *Human Evolution*, Chicago, 1970, Aldine.
- Cohen MM, Jr: *The Child with Multiple Birth Defects*, ed 2, New York, 1997, Oxford University Press.
- Cohen MM, Jr, MacClean RE: *Craniosynostosis: Diagnosis, Evaluation, and Management*, New York, 2000, Oxford University Press.
- Cohen MM, Jr: Perspectives on Craniofacial Anomalies, Syndromes, and Other Disorders. In Lin KY, Ogle RC, Jane JA, editors: *Craniofacial Surgery: Science and Surgical Technique*, Philadelphia, 2002, W.B. Saunders Company, Chapter 1, pp. 3–38.
- Cohen MM, Jr: FGFs/FGFRs and Associated Disorders. In Epstein CJ, Erickson RP, Wynshaw-Boris A, editors: *Inborn Errors of Development*, 2004. New York, 2004, Oxford University Press, Chapter 33, pp 380–400.
- Cohen MM, Jr: *Perspectives on the Face*, New York, 2006, Oxford University Press.
- Cohen MM, Jr: Craniofacial Disorders. In Rimoin DL, Conner JM, Pyeritz RE, Korf BR, editors: *Emory and Rimoin's Principles and Practice of Medical Genetics*, ed 5, Philadelphia, 2007, Elsevier, Chapter 151, pp 3303–3348.

12. Cohen MM, Jr: Craniofacial Abnormalities. In *Potter's Pathology of the Fetus, Infant, and Child*, ed 2, Philadelphia, 2007, Elsevier, Chapter 20, pp 885–918.
13. Cohen MM, Jr: *Mental retardation, psychiatric disorders, neurodegenerative disorders, and genetics*. PowerPoint presentation dedicated to my Fellow Angela M. Castro, MD. 2010.
14. Doubilet D: *Fishface*, New York, 2003, Phaidon Press.
15. Elffers J: *Play with Your Food*, New York, 1997, Stewart, Tabori & Chang.
16. Enlow DH: *The Human Face*, New York, 1968, Harper & Row.
17. Enlow DH: *Facial Growth*, ed 3, Philadelphia, 1990, Saunders.
18. Evans PD, Gilbert SL, Mekel-Bobrov N, Vallender EJ, Anderson JR, Vaez-Azizi LM, Tishkoff SA, Hudson RR, Lahn BT: *Microcephalin*, a gene regulating brain size, continues to evolve adaptively in humans. *Science* 309:1717–1720, 2005.
19. Ezhkova E, Fuchs E: An eye to treating blindness. *Nature* 466:567–568, 2010.
20. Fagerholm P, Lagali NS, Merrett K, Jackson WB, Munger R, Liu Y, Polarek JW, Söderqvist M, Griffith M: A biosynthetic alternative to human donor tissue for inducing corneal regeneration: 24-month follow-up of a phase I clinical study. *Sci Transl Med* 2:46ra61, 2010.
21. Geschwind N: Specialization of the human brain. In *The Brain: A Scientific American Book*, San Francisco, 1979, W.H. Freeman and Co., pp 108–117.
22. Gilbert CA: *All Is Vanity*. New York: House of Art.
23. Gould SJ: *Ontogeny and Phylogeny*, Cambridge, UK, 1977, Harvard University Press.
24. Hayakawa T, Angata T, Lewis AL, Mikkelsen TS, Varki NM, Varki A: A human-specific gene in microglia. *Science* 309:1693, 2005.
25. Jirásek JE: *Atlas of Human Prenatal Morphogenesis*, Boston, 1983, Martinus Nijhoff.
26. Kenerknecht I, Grueter T, Welling B, Wentzek S, Horst J, Edwards S, Gueter M: First report of prevalence of non-syndromic hereditary prosopagnosia (HPA). *Am J Med Genet* 140A:1617–1622.27, 2006.
27. Klein RG: Whither the Neanderthals? *Science* 299:1525–1527, 2003.
28. Krings M, Capelli C, Tschentdcher F, Geisert H, Meyer S, von Haeseler A, Grossschmidt K, Possnert G, Paunovic M, Pääbo S: A view of Neanderthal genetic diversity. *Nat Genet* 26:144–146, 2004.
29. MacLaren RE, Pearson RA, MacNeil A, Douglas RH, Salt TE, Akimoto M, Swaroop A, Sowden JC, Ali RR: Retinal repair by transplantation of photoreceptor precursors. *Nature* 444:203–207, 2006.
30. Maguire AM, Simonelli F, Pierce EA, Pugh EN, Mingozzi F, Benicelli J, Banfi S, Marshall KA, Tesa F, Surace EM: Safety and efficiency of gene transfer for Leber congenital amaurosis. *New Engl J Med* 358:2240–2248, 2008.
31. Mekel-Bobrov N, Gilbert SL, Evans PD, Vallender EJ, Anderson JR, Hudson RR, Tishkoff SA, Lahn BT: Ongoing adaptive evolution of *ASPM*, a brain size determinant in *Homo sapiens*. *Science* 309:1720, 2005.
32. Moffett BC, Jr: *Syllabus for course in fundamentals of skeletal biology related to clinical dentistry*. Unpublished manuscript, 1973. University of Washington.
33. Parr LA, Heintz M: Rhesus monkey expressions. *Anim Behav* 77:1507–1513, 2009.
34. Pruzansky S: Clinical investigation of the experiments of nature. *ASHA Report* 8:62–94, 1973.
35. Radinsky LB: *The Evolution of Vertebrate Design*, Chicago, 1987, University of Chicago Press.
36. Rama P, Matuska S, Paganoni G, Spinelli A, De Luca M, Pellegrini G: Limbal stem-cell therapy and long-term corneal regeneration. *New Engl J Med* 363:147–155, 2010.
37. Scammon R: *Section 1 in Human Anatomy by H. Morris*, ed 11, Philadelphia, 1953, Blakiston.
38. Sharma RK: Developmental expression of GABA_A receptors in retinal transplants. *Ophthalmic Res* 32:45–51, 2000.
39. Tassabehji M, Hammond P, Karmiloff-Smith A, Thompson P, Thorgerisson SS, Durkin ME, Popescu NC, Hutton T, Read AP, Macconochie M, Donnai D: *GTF2IRD1* in craniofacial development of humans and mice. *Science* 310:1184–1187, 2005.
40. Thumann G: Nonviral gene therapy for age-related macular degeneration. *Expert Rev Ophthalmol* 6:81–93, 2011.
41. Tibbetts EA, Dale J: A socially enforced signal of quality in a paper wasp. *Nature* 432:218–222, 2004.
42. Witten PE, Hall BK: Seasonal changes in the lower jaw skeleton in male Atlantic salmon (*Salmo salar* L.): Remodeling and regression of the kype after spawning. *J Anat* 203:435–450, 2003.

2

Pioneers and Milestones in the Field of Orthodontics and Orthognathic Surgery

JEFFREY C. POSNICK, DMD, MD

- [Pioneers in the Field of Orthodontics](#)
- [Early Pioneers in the Field of Orthognathic Surgery](#)
- [Hugo L. Obwegeser: Development of the Standard Orthognathic Procedures](#)
- [William H. Bell: Experimental Studies to Confirm the Biologic Basis of the Standard Orthognathic Procedures](#)
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Pioneers in the Field of Orthodontics

Background

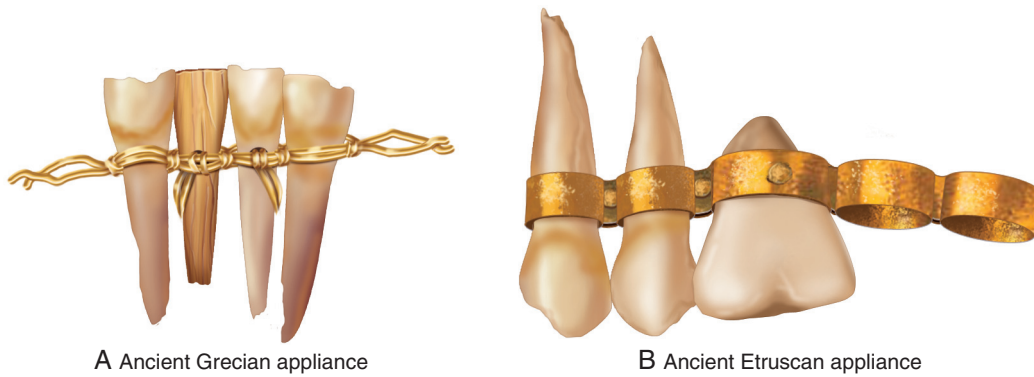
Orthodontic appliances that are somewhat primitive but surprisingly well-designed for their time have been found in Ancient Greek written materials.⁸ Four Etruscan specimens of young adult women dating back to 900 B.C. that specifically demonstrate appliances on the teeth have also been recovered.^{23,208} The orthodontic type appliances are gold bands around teeth that are adjacent to an edentulous gap. The appliances appear to be for the purpose of managing the space left by tooth loss earlier in life (Fig. 2-1).²⁰⁹ In addition, the Ancient Greek scholars Hippocrates and Aristotle both have writings that discuss various ways to straighten teeth and to treat various dental conditions.^{20,51} Knowledge of dental extractions carried out for the purpose of mechanically straightening the remaining teeth was also reported by Leonardo da Vinci during the High Italian Renaissance.¹⁶⁶ The bandolet or bandeau, which was created in 1723 by Pierre Fauchard of France, was the first documented dental

arch expansion appliance (Fig. 2-2).²⁰² It consisted of a heavy maxillary labial arch wire to which the teeth were ligated.²⁰² The term *orthodontia* is said to have been coined in 1841 by Lafoulon, and it appeared in the book about malocclusion written by J.M. Alexis Schange.^{11,157,203}

In 1858, perhaps the first formal article on orthodontics published in a medical journal was written by Norman W. Kingsley (Fig. 2-3). By the 1880s, Norman Kingsley's *Treaties on Oral Deformities as a Branch of Mechanical Surgery* systematically described the current techniques of the day for the repositioning of teeth (Fig. 2-4).¹⁰⁹ Kingsley was among the first to fully describe how extraoral forces (e.g., headgear) could be used to correct protruding maxillary anterior teeth. His emphasis was primarily on the alignment of the anterior maxillary teeth in an attempt to improve facial appearance. To Kingsley, the extraction of teeth for the purpose of uncrowding and to reduce the protraction of the incisors was acceptable practice. Details of how the upper and lower teeth articulated with each other (i.e., occlusion) were of secondary importance. He used a wide variety of apparatus to reposition the anterior teeth, including wires, bands, custom palatal plates, elastics, wedges, linen twine, rubber tubing, leathers, gold bands, and vulcanite. Impressions of the teeth and dental models were used during planning and for the precontouring of labial placed wires. The use of an external apparatus placed over the top of the head and then either secured to the chin or held with bands secured to the maxillary incisors became part of his everyday practice (Fig. 2-5).

Kingsley's basic principles for the mechanical alignment of the teeth can be summarized as follows:

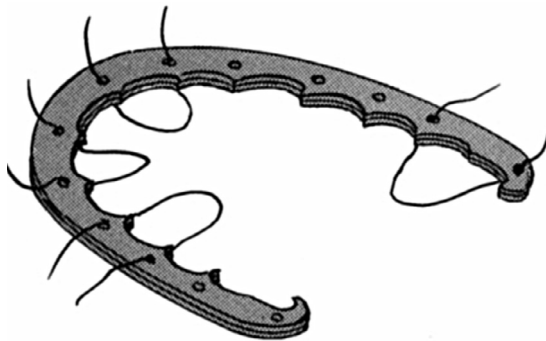
- The positioning of the six maxillary anterior teeth with regard to each other and in relationship to the lip is essential for facial aesthetics.
- The contact of a certain number of maxillary and mandibular posterior teeth, at least on one side, is important for the grinding and mastication of food.



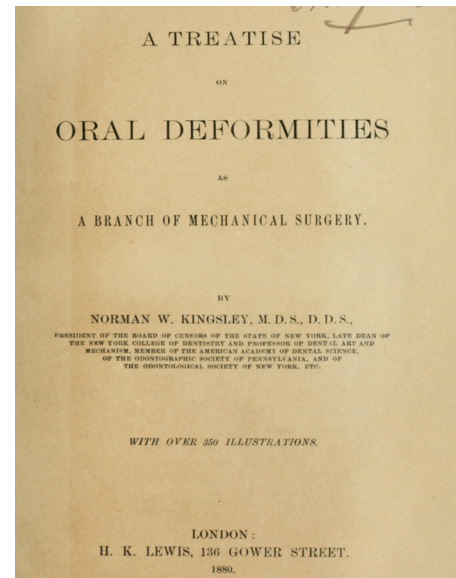
A Ancient Grecian appliance

B Ancient Etruscan appliance

• **Figure 2-1 A**, A drawing of the anterior mandibular dentition in a mummy specimen from ancient Greece (approximately 1000 B.C.) currently housed in the Archaeological Museum in Athens, Greece. A gold wire was used to tie teeth together and to band others to support replacements. The use of human, animal, carved wood, bone, or ivory for replacement teeth was practiced by the ancient Grecian society. **B**, A drawing of an ancient Etruscan orthodontic appliance found on the maxilla of a mummy in a tomb (approximately 700 B.C.), which is currently at the Civic Museum in Cornet, Italy. It demonstrates gold soldered rings and rivets to hold dental replacements as a bridge. The specimen has two natural teeth, one riveted oxtooth and four spaces for others



• **Figure 2-2** Illustration of the “bandolet” bandeau, which was created in 1723 by Pierre Fauchard of France. This was the first documented dental arch expansion appliance.



• **Figure 2-4** Norman Kingsley's text entitled *A Treatise on Oral Deformities as a Branch of Mechanical Surgery*, which was published in 1880.

- All other teeth are potentially expendable, depending on the effort required by the clinician for straightening and the individual's desire to maintain them.
- From both a mastication and an aesthetic perspective, there is no particular need for the maintenance of all 32 teeth or to achieve a specific and detailed way of the upper and lower teeth occluding with one another.

Dr. Kingsley also commented on facial aesthetics, stating that “the eye soon tires of the stiffness and formality of unbroken uniformity, and is only permanently pleased with the beauty which comes from graceful variation.”¹⁰⁹ He acknowledged the occurrence of jaw deformities when he said that “[a] lack of harmony between the maxilla and mandible is occasionally seen. For instance, when the



• **Figure 2-3** Photograph of Norman W. Kingsley (1829-1913).



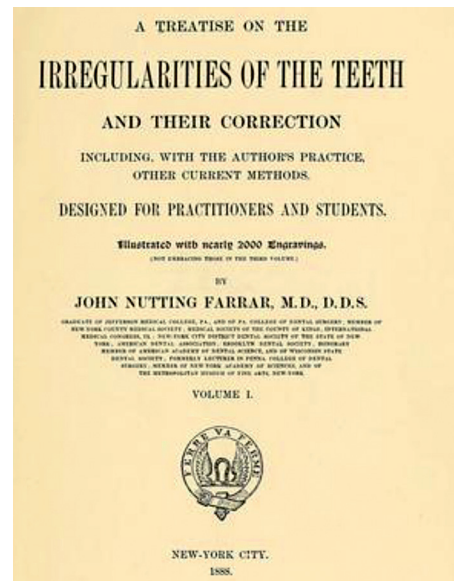
• **Figure 2-5** Headgear designed and used by Norman W. Kingsley. According to Kingsley, "I sat to work to make an apparatus that would pull the lower jaw back. As you can see from the photograph taken at the time she was wearing the apparatus, it consists of two parts. For the lower part, I made a brass plate to fit the chin, having arms with hooked ends reaching to a point just below the point of the chin. These arms were arranged in such a way that the distance between them could be altered at will by simply pressing them apart or to gather. The upper part consisted of a simple network going over the head and having two hooks on each side, one hook being above and the other below the ear. When the apparatus was completed and in use, there were four ligatures of ordinary elastic rubber pulling in such a way as to force the lower jaw almost directly backward." *From Kingsley NW: A treatise on oral deformities as a branch of mechanical surgery, London, 1880, H.K. Lewis, pp 137, Figure 68.*

mandible is very large and massive or unusually small in comparison to the upper jaw. When present, this will interfere with the harmony of the surrounding parts of the face."¹⁰⁹

In 1897, J.N. Farrar published a two-volume illustrated text entitled *A Treatise on the Irregularities of the Teeth and Their Corrections* (Fig. 2-6).¹² In that text, he also demonstrates detailed orthodontic appliances, and he was among the first to suggest the advantages of mild (rather than heavy) force applied at intervals to effectively move the teeth.¹⁰

Edward H. Angle

It was common practice through the later half of the 19th century to extract the teeth in response to most dental problems.^{9,60} Restorative dentistry was generally not the first choice for the management of "toothaches." Most adults at that time (i.e., the 1880s) did not have a full complement of teeth nor was there much stigma associated with partial edentulism.²¹⁰ Details of how the teeth occluded with each other seemed important only when establishing how upper and lower dentures were to meet so as to prevent loosening or mobility when wearing them and to improve chewing ability. It was a natural extension of this thinking for a



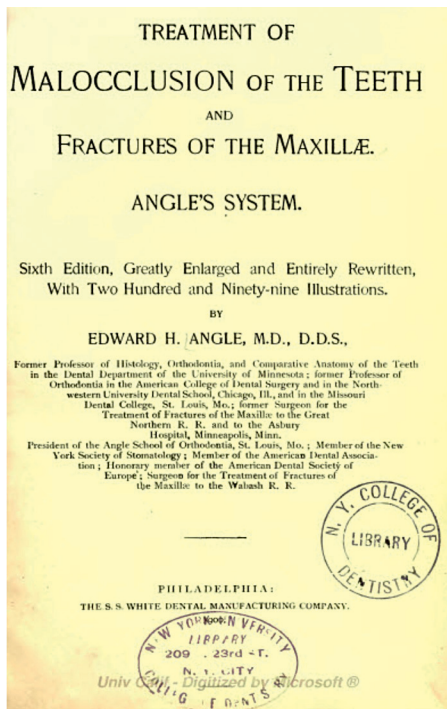
• **Figure 2-6** John Nutting Farrar's two-volume illustrated text entitled *A Treatise on the Irregularities of the Teeth and Their Correction*, which was published in 1897.



• **Figure 2-7** Photograph of Edward H. Angle (1855-1930).

prosthodontic-trained dentist like Edward H. Angle to develop a more holistic concept regarding the importance of the occlusion of the natural dentition (Fig. 2-7).⁵⁻⁸ During his career, Angle taught prosthetics at the University of Minnesota (Minneapolis, Minn), Northwestern University Dental School (Chicago, Ill), and the Missouri Dental College (St. Louis, Mo). Dr. Angle's interest in dental occlusion and his ingenuity and dedication with regard to the correction of malocclusion led to the development of the specialty that we now call "orthodontics."

Angle's textbook, *Treatment of Malocclusion of the Teeth*, was first published in 1887 (Fig. 2-8).⁶ It went into seven much-revised editions, and it laid the foundation for the modern specialty of orthodontics. The publication of Angle's classification of malocclusion during the 1890s was a key step in the development of orthodontics (Fig. 2-9).^{42,43}



• **Figure 2-8** Dr. Angle published a comprehensive text entitled *A Treatment of Malocclusion of the Teeth and Fractures of the Maxillæ*, the first edition of which was published in 1887. It went into seven revised editions, and it laid the foundation for the modern specialty of orthodontics.

Angle introduced the first precise and simple definition of the normal human occlusion of the natural dentition. He postulated that a key aspect of how the teeth should fit together was the positioning of the maxillary first molars. He then went on to describe three classes of malocclusion that were based on the occlusal relationships of the first molars:

Class I: There is a normal relationship of the molars, but the line of occlusion is incorrect as a result of malposed teeth, rotations, or other causes.

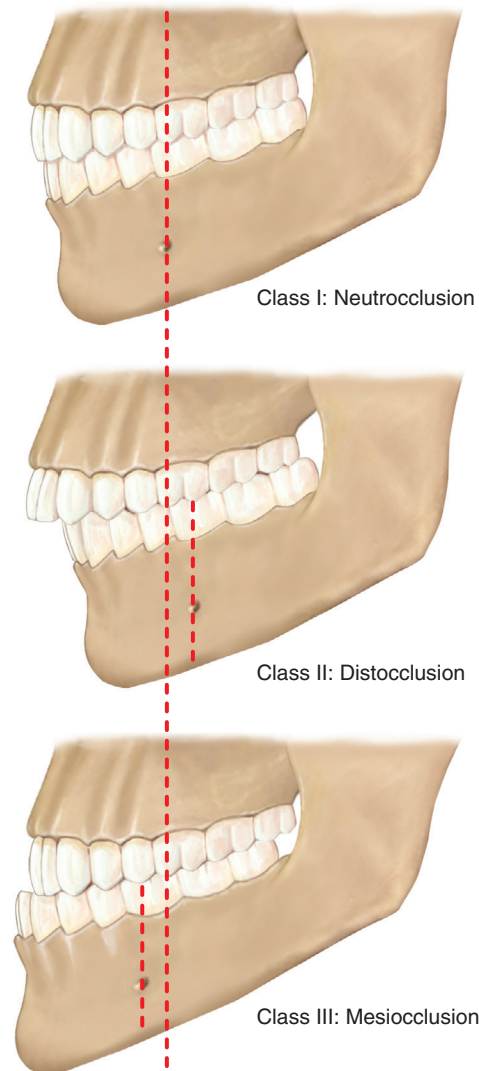
Class II: The lower molar is distally positioned relative to the upper molar, and the line of occlusion is not specified.

Class III: The lower molar is mesially positioned relative to the upper molar, and the line of occlusion is not specified.

Angle's classification actually has four classes:

- Normal occlusion (Class I, with a normal line of occlusion)
- Class I malocclusion
- Class II malocclusion
- Class III malocclusion

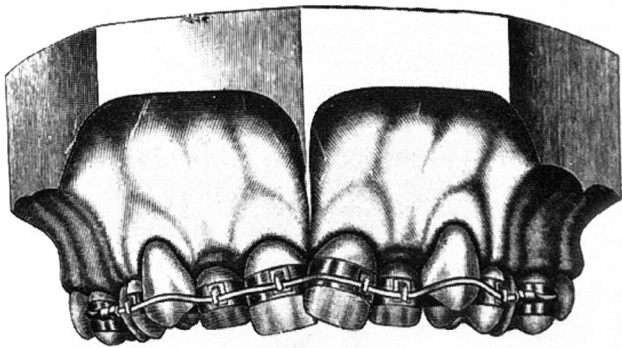
Normal occlusion and Class I malocclusion share the same molar relationship, but they differ with regard to the arrangement of the other teeth relative to the line of



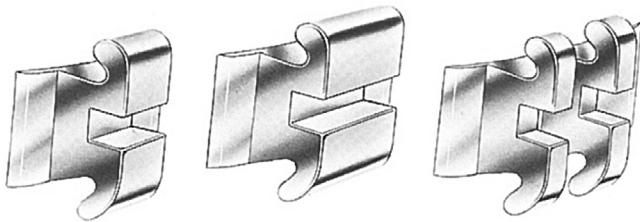
• **Figure 2-9** Angle described three classes of malocclusion based on the relationships of the first molars: Class I, Class II, and Class III.

occlusion. The line of occlusion may or may not be correct in Class II and Class III.

By the early 1900s, orthodontists no longer just aligned irregular teeth in each arch.^{175,218} The practice had now evolved into the treatment of malocclusion (i.e., how the upper and lower teeth align with each other). Angle's philosophy was to achieve a specific relationship between the upper and lower teeth for each patient. Since Angle's precisely defined dental relationships also required a full complement of teeth in each arch, the maintenance of an intact dentition became an important component of orthodontic treatment. Angle and his followers strongly opposed extraction for orthodontic purposes.⁶⁹ In an attempt to achieve the three-axis control of tooth movement, Angle introduced the *ribbon arch* (Fig. 2-10).⁷⁰ He then devised the *edgewise appliance* to overcome the weaknesses of the ribbon arch (Fig. 2-11). The Angle reoriented bracket slot was designed to control the bodily movement of teeth and to limit tipping.¹³⁵ Angle also heavily relied on the use of intraoral



• **Figure 2-10** The ribbon arch appliance, which was introduced by Angle in 1916. The vertical slot in the bracket accepts either round or rectangular wire, which in turn is held in place by a locked pin.



• **Figure 2-11** The edgewise bracket attachment comes in a variety of modifications. The original single bracket (*left*) has been widened for molar teeth and for greater rotational control (*center*). The twin edgewise modification (*right*) is available in different widths, and it is used most frequently because of its greater versatility, its reduced frictional component, and its adaptability to various light-wire techniques.

rubber bands and limited the use of extraoral appliances (e.g., headgear) as much as possible.

Angle was among the first to state that jaw discrepancies exist in some individuals and that these discrepancies will prevent the achievement of a normal occlusion. He then acknowledged the need for jaw-straightening surgery. In Chapter 14: Operative Surgery of the sixth edition of Angle's textbook, he states that the use of appliances to move teeth may be properly called *conservative surgery* to distinguish it from more bold or aggressive operations involving the use of cutting instruments, which were designated as *operative surgery*. He states the following: "While such operations [mandibular osteotomies] should probably be employed only as auxiliary to the conservative method, they are doubtless destined to play an more important part in the practice in the future."⁶

Dr. Angle worried that no technique that relied solely on tooth movement could establish the appropriate relationships of the teeth or truly improve the facial lines in cases involving the severe overdevelopment of the inferior maxilla [mandible].⁶ He felt that these cases may be successfully treated by removing a section of bone from each of the lateral halves of the mandible. At the time, the removal of a single complete section of the jaw had been reported for cases involving such conditions as ankylosis, tumors, and gunshot wounds, but Dr. Angle was unable to find information about the removal of complete sections from each of

the lateral halves of the mandible. This idea was discussed by Dr. Angle with various surgeons and dentists and determined to be feasible. His proposal involved the following⁶:

- Careful photographs should first be taken of the patient.
- Two accurate models should be made of the lower dental arch, and one should be made of the upper.
- One of the plaster models of the lower jaw should then be sawed through and the sections removed.
- The positions and extent of the sections should be carefully experimented with until the three remaining sections of the model could be made to best harmonize with the upper arch so that the teeth may be in best possible occlusion with those of the upper jaw.
- The sections of the plaster model should then be cemented or waxed together.
- Over this reconstructed model, a vulcanite or metal splint should then be formed.
- With the use of careful comparisons and measurements of the reconstructed model as compared with the unchanged model, the exact size and form of both resections of bone to be removed can be determined so that there need be no guessing as to the relationships of the bone. In this way, complete apposition of the cut ends can be made.
- Because there is a lingual inclination of the lower incisors in all cases of mandibular prognathism, the sections of bone to be removed must not be parallel on their sides but instead more or less wedged or V-shaped to gain the best positions for the occlusion of the incisors as well as for the appearance of the chin.
- The degree of variation from the parallel of the lines of bone resection must be determined by the conditions that are present (i.e., patient variation).
- Because the operation must be skillfully performed, the most rigid support should be given to the reconstructed jaw. This technique (i.e., a cap splint over the teeth) should give more rigid support than is possible with the crude, unstable, and unmechanical plan of wiring the ends of the bone together, which is so often employed for the reduction of fractures.

According to Dr. Angle, "The question most often raised by dentists in discussing the practicability of an operation as outlined above was the uncertainty as to union of the bones and as to impairment of vitality of the teeth in the anterior segment [of the mandible]."³⁶ However, Dr. Angle helped with the overcoming of these concerns via two case reports that were presented in his textbook, *Treatment of Malocclusion of the Teeth and Fractures of the Mandible: Angle's System*⁶ (pp 173-184):

First Patient (Operation)

June 23, 1897, the author [Dr. Angle] assisted the late Professor Henry H. Mudd in one of these operations performed at St. Luke's Hospital, St. Louis, upon Ms. M.J. of Arkansas, a delicate girl, 13 years old. She had likely suffered with generalized sepsis at 3 years of age

with the mandible becoming fixed and firmly closed [temporomandibular joint ankylosis]. The upper jaw was normal in size and contained a full complement of teeth. The lower jaw was deformed. The ramus of the jaw on each side passed downward and backwards so that the angle of the jaw came a little behind rather than in front of the vertical line dropped from the lobe of the ear. The arch of the lower jaw was broad and the incisor teeth were impinging the surface of the palate. The only point of entrance for food was the opening under the arch of the hard palate above the incisor teeth of the lower jaw.

To free the body of the jaw so that the mouth might be opened, a triangular segment with the base downward and the apex upward and forward was removed from the angle of the jaw on each side. A complete section of the bone was thus removed on each side to make false joints at the junction of the ramus and the body of the mandible. When the bone was separated, the tongue and jaw dropped down, making respiration difficult. An urgent tracheostomy was made. Wire ligatures were used to establish IMF [intermaxillary fixation]. This pulled the tongue and neck muscles superior and anterior, and established a good airway. The ligatures were occasionally removed in the ensuing weeks to allow movement of the jaw and establish a hinge joint instead of osseous union in the ramus regions of the mandible. Unfortunately, the patient was lost to follow-up several weeks later without permanent record of how things turned out.

Second Patient (Operation)

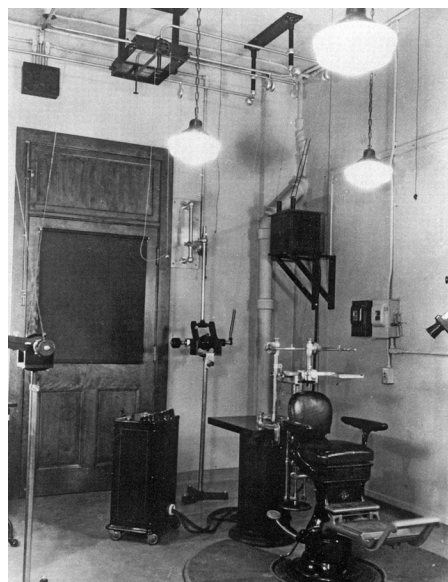
A double resection [of the mandible] for the purpose of shortening a prognathic mandible was performed successfully at the Baptist Hospital in St. Louis [Dr. Vilray Blair was the surgeon, and the procedure was performed on December 19, 1897]. Such an operation might at first seem formidable, but there is no reason why, if skillfully performed according to modern aseptic methods of surgery and the plan is indicated, that clean smooth ends of the bone should not unite at least as readily as they do in the common cases of double [mandibular] fractures and the result being nearly ideal.

Holly Broadbent

The introduction of the cephalometer by Holly Broadbent in 1931 was another important milestone that placed orthodontic research on a scientific foundation (Fig. 2-12).¹⁵⁷ For the first time, it made possible the accurate study of the facial bones in the growing child (Figures 2-13 and 2-14). It became a valuable supplement to the orthodontist's plaster models and intraoral x-rays. The value of serial cephalograms in clinical practice was quickly realized. Broadbent's original work on the "face of the normal child" and Brody's classic research "on the growth pattern of the human head from the third month to the eighth year of life" were among the earliest contributions.¹⁶³ These were followed by the research of Down's "Variations in Facial Relationships,"

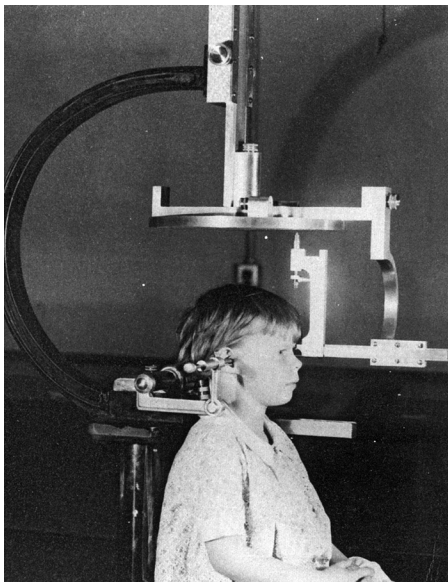


• **Figure 2-12** Photograph of Holly Broadbent (1894-1977). From Broadbent BH Sr., Broadbent BH Jr and Golden WH: *Bolton Standards of Dentofacial Developmental Growth*, St. Louis, C.V. Mosby, 1975.



• **Figure 2-13** Broadbent was the first to develop the standardized lateral "head plate." The cephalometer is a device for holding the patient's head, the x-ray film, and the central ray of the x-ray machine in proper relationship with one another. Ear-rod extensions are necessary to adjust the head so that the profile is centered, regardless of the size or shape of the head. The orbital pointer and the ear rods adjust the patient's head along the horizontal or Frankfort plane. An adjustable chair is used, and the teeth are placed in "centric occlusion" unless otherwise indicated. The cephalometer may be rotated so that posterior and anterior radiographs as well as profile radiographs can be taken. The central ray is directed through the ear rods, thereby producing a circle on the x-ray film. From Broadbent BH Sr., Broadbent BH Jr and Golden WH: *Bolton Standards of Dentofacial Developmental Growth*, St. Louis, C.V. Mosby, 1975.

the work of Thompson in "Functional Analysis of Occlusion," Wylie's "Assessment of Anterior-Posterior Dysplasia," and the work of Margolis' "Basic Facial Pattern and Its Application in Clinical Orthodontics."¹⁷⁴ The use of cephalometric radiography in clinical practice became widespread



• **Figure 2-14** The cephalostat device, as described in [Figure 2-13](#), is shown with a patient who is positioned and ready for film exposure. From Broadbent BH Sr., Broadbent BH Jr and Golden WH: *Bolton Standards of Dentofacial Developmental Growth*, St. Louis, C.V. Mosby, 1975.

after World War II and made clear to all clinicians that many of the Class II and Class III malocclusions being treated actually resulted from abnormal skeletal relationships rather than just malposed teeth.¹⁶⁵ These radiographic truths were hard to ignore and further inspired orthodontists and surgeons to search for operative solutions.¹⁸¹

Charles Tweed and Raymond Begg

Another “orthodontic truth” was also soon to fall. A purest “Angle” approach to maintaining and straightening all of the teeth in all patients frequently proved untenable with regard to the maintenance of the occlusion after the appliances were removed.^{54,144,145} This non-extraction, arch-expansion approach gave little consideration to long-term periodontal health or to the individual’s overall facial aesthetics.¹⁶¹ The extraction of teeth (typically the bicuspid) to achieve occlusal stability and periodontal health was reintroduced into American orthodontics during the 1930s by Charles Tweed ([Fig. 2-15](#)) and simultaneously into the United Kingdom (Australia) by Raymond Begg ([Fig. 2-16](#)) (see Chapter 17 for in-depth discussion).^{47-49,59,204}

Lawrence F. Andrews

Dr. Lawrence F. Andrews’ pioneer contributions to the field of orthodontics and the treatment of dentofacial deformity must also be mentioned ([Fig. 2-17](#)).²⁻⁴ After completing orthodontics training at Ohio State University in 1958, he entered private practice that was limited to orthodontics. Early in his career, Dr. Andrews recognized clinical problems that were yet unsolved and sought solutions. This sent him



• **Figure 2-15** Photograph of Charles Tweed (1895-1970).



• **Figure 2-16** Photograph of Raymond Begg (1889-1983). From Wahl N: *Orthodontics in 3 millennia. Chapter 6: More early 20th-century appliances and the extraction controversy*. *Am J Orthod Dentofacial Orthop* 128:795-800, 2005.



• **Figure 2-17** Photograph of Lawrence F. Andrews.

on a search to better understand occlusion as well as deviations from normal. Between 1960 and 1964, Dr. Andrews began to gather data for analysis. One hundred and twenty non-orthodontic normal (dental) models were acquired with the cooperation of local dentists, orthodontists, and a major university. The models selected for study were of teeth that had never undergone orthodontic treatment; that were straight and pleasing in appearance; that had a bite that looked generally correct; and that, in Dr. Andrews' judgment, would not benefit from orthodontic treatment.

Dr. Andrews studied the crowns of this multisource collection of models to ascertain which characteristics, if any, would be found consistently in all of them. Angle's molar cusp groove concept was validated but found to be incomplete. After meticulous evaluation, including comparison with "treated cases" from the nation's most skilled orthodontists (n = 1150), six consistent occlusal characteristics were formulated by Dr. Andrews in general terms and then explained in detail.² The six fundamental qualities or *Six Keys of Optimal Occlusion* were validated not solely because all were present in each of the non-orthodontic normal models (n = 120) but because the lack of even one of the six was a defect that was predictive of an incomplete end result in the orthodontically treated models that were studied (n = 1150).

The significant characteristics (i.e., the *Six Keys of Optimal Occlusion*) described by Andrews that were found in all of the non-orthodontic normal models were as follows:

- Key I: Interarch (molar) relationship
- Key II: Crown angulation
- Key III: Crown inclination
- Key IV: Rotations
- Key V: Tight contacts
- Key VI: Curve of Spee

In his published article, Dr. Andrews acknowledged that, although each normal person is unique in his or her own way, they nevertheless have much in common.² The 120 non-orthodontic normal models studied differed with regard to certain aspects, but all shared the Six Keys. Dr. Andrews suggested that we use as our benchmark "nature's best" and that, in the absence of abnormalities outside of our control, we limit compromise and strive for the ideal. He developed the *straight-wire appliance* in 1970 for orthodontic use to help accomplish these objectives. This preadjusted appliance soon became the standard of the specialty.²⁻⁴

Dr. Andrews then went on to describe six fundamental principles that are necessary to achieve both facial and dental harmony: the *Six Elements of Orofacial Harmony*. Both of these aspects—facial and dental harmony—are frequently absent in the patient with dentofacial deformity. Dr. Andrews was one of the first orthodontist to clarify the importance of addressing both facial and dental harmony from the beginning of treatment to achieve the best facial form and head and neck function for each patient. Dr. Andrews stated that, for the maxillomandibular complex to

be in harmony (i.e., to have all elements in alignment) with the overall face, the following must be present:

- Element I: Proper arch shape and positioning of the maxillary and mandibular teeth (roots) over the basal bone
- Element II: Proper horizontal (sagittal) projection of the maxilla
- Element III: Proper width of the maxillary and mandibular arches
- Element IV: Proper vertical height of the maxilla
- Element V: Proper prominence (shape) of the chin (i.e., pogonion prominence)
- Element VI: Establishment of the *Six Keys to Optimal Occlusion*

He studied each of these Six Elements and defined both qualitatively and quantitatively how the orthodontist and the surgeon can work together to achieve these objectives for each patient.

William R. Proffit

By the mid 1970s, Dr. Proffit recognized the benefits of a collaborative surgeon-orthodontist interaction for the correction of dentofacial deformities.¹⁶⁴ He and others convincingly demonstrated to surgeons that rectangular orthodontic arch wires in edgewise brackets were satisfactory for surgical patients without the need for the intraoperative placement of Erich arch bars. This paved the way for the routine preoperative orthodontic relief of dental compensations, and it was followed by the placement of orthodontic "surgical wires and hooks" for intraoperative use. After the initial surgical healing, a smooth transition to orthodontic maintenance and detailing soon became the standard of care. Dr. Proffit was also an early believer in the importance of a collaborative effort to understand the patient's presenting dysfunction and dysmorphology and then to develop a comprehensive treatment plan. The use of a cephalometric analysis with prediction tracings followed by dental model planning with splint construction became the routine. Dr. Proffit and others also insisted on analysis of the early results that were achieved. This was followed by critical studies of the long-term outcomes after various surgical approaches and orthodontic techniques were employed. This analytic approach has been responsible for much progress in the field over the past four decades.

According to Dr. William R. Proffit* (Fig. 2-18), orthodontics in the 21st century differs from that of the previous century in three fundamental ways^{162,163}:

1. *There is more emphasis on both facial and dental aesthetics and less on the details of dental occlusion as an overriding objective.* This has much to do with the safe and reliable orthognathic surgical techniques that are currently

*Please note that the author of this text has paraphrased Dr. Proffit's original work.



• **Figure 2-18** Photograph of William R. Proffit (1936–Present)

available and with the recognition that long-term stable occlusion and periodontal health can best be achieved in conjunction with harmonious jaw relationships. This will require extractions when necessary to uncrowd and achieve periodontal health; decompensating orthodontic therapy to place teeth that are solidly in the alveolar bone; and jaw-straightening surgery, when indicated.

2. *Patients and families expect greater involvement in the treatment planning process.* Full disclosure requires the orthodontist to consider and review ideal and compromised treatment options. Providing a balanced and objective assessment of each option's effect on facial aesthetics, dental stability, periodontal health, speech articulation, and the upper airway is ideal. Consultation with appropriate dental, surgical, and medical specialists before instituting treatment has become the standard of care.
3. *Orthodontics is now offered not just to children, adolescents, and young adults but also to older adults through an interdisciplinary approach.* Consideration should be given to facial aesthetics, occlusion, periodontal health, the upper airway, speech articulation, and the long-term maintenance of the dentition. There is increased emphasis on coordinated treatment with other dentists, surgeons, and medical specialists. Taking advantage of currently available sophisticated orthodontics and orthognathic surgery to provide individuals of all ages with quality care should now be considered routine.

Early Pioneers in the Field of Orthognathic Surgery

Mandibular Setback for Prognathism

Simon Hüllihen described a procedure for the correction of mandibular dentoalveolar protrusion in the *American Journal of Dental Science* in January 1849 (Fig. 2-19, A).^{18,98} Technically, this was a bilateral bicuspid region wedge osteotomy to “set back” the anterior mandibular dentoalveolar segment. The osteotomy did not violate the inferior border

of the mandible. The procedure was performed on a middle-aged woman whose mandibular deformity was the result of a severe burn scar contracture of the anterior neck and lip from an injury that had occurred during her childhood (see Fig. 2-19, B and C).

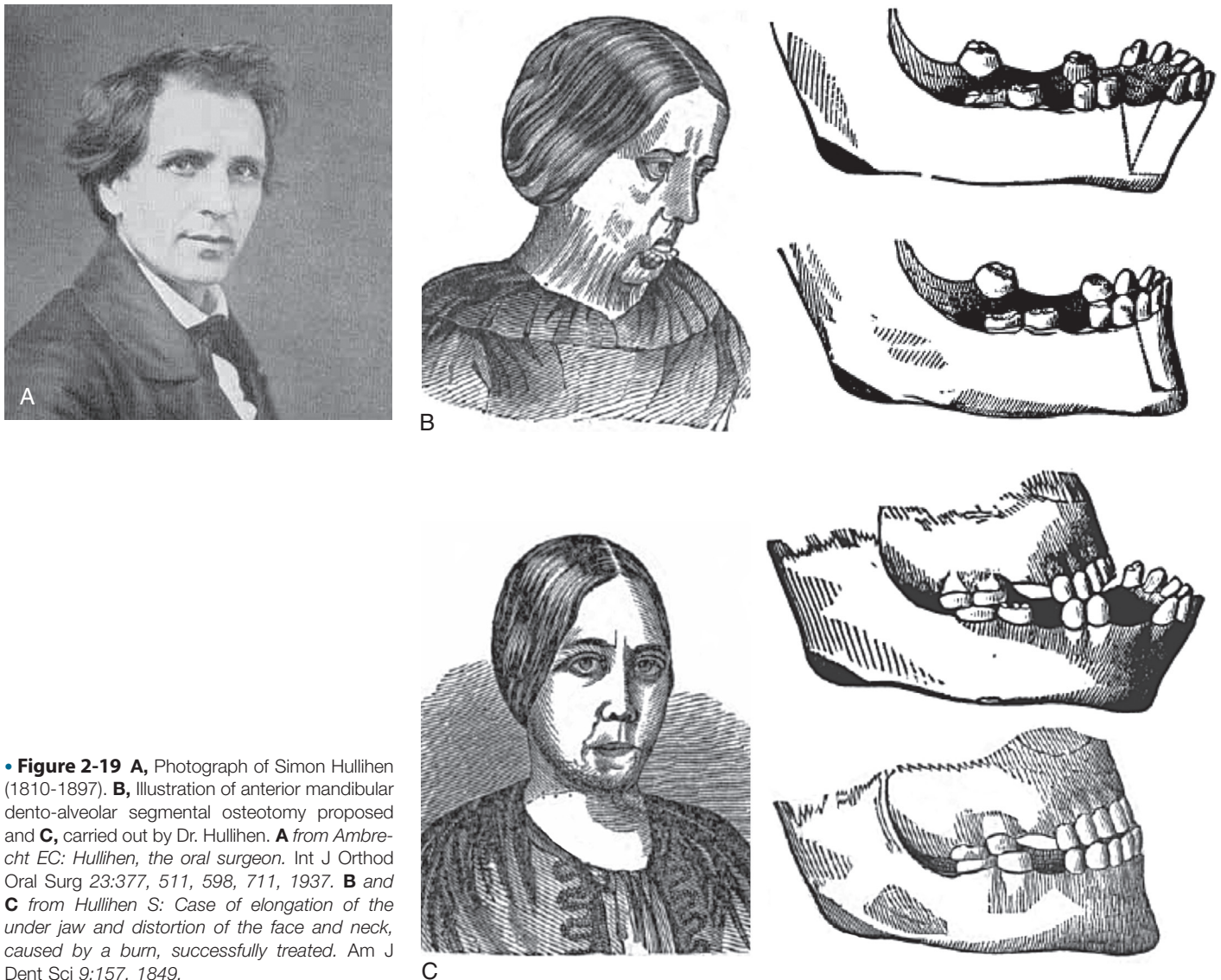
In 1887, Berger from Lyon, France, described bilateral condylar neck osteotomies to set back the prognathic mandible.⁹⁰ In 1895, Jaboulay also reported the bilateral osteotomy of the condylar neck (condylectomy) for the correction of prognathism.¹⁰⁰ This method continued to be popular in France for the treatment of prognathism through the 1950s.⁷⁵

On December 19, 1897, Vilray Blair in St. Louis, Mo, carried out a distinct modification of the original Hüllihen procedure for the treatment of mandibular prognathism (see the “Second Patient (Operation)” case report by Dr. Angle earlier in this chapter) (Fig. 2-20).³⁸ The operation was performed on a 22-year-old man with asymmetric mandibular prognathism. The young man had difficulty with chewing and speech articulation, and he also was self-conscious about his appearance; thus, he was enthusiastic about proceeding with the recommended surgical plan. Dr. Blair stated that the immediate surgical objective was to carry out bilateral body osteotomies to shorten the horizontal ramus [body of the mandible] and to change the mandibular angle [rotate the distal mandible counterclockwise], thereby allowing for the closure of the open bite. Dr. Blair's preoperative concerns about the operation included the following:

1. Could a portion of the mandible be cut out on each side in a satisfactory way?
2. Would sacrifice of the inferior dental nerve cause detrimental injury to the distal mandible?
3. After healing had occurred, would the mandible continue to grow and cause a recurrence of the deformity?

Through cadaver dissections, Dr. Blair satisfied his concerns regarding whether the mandible could be sectioned and repositioned successfully. As a result of the age of the patient (22 years), he had little concern regarding further growth after surgery. He conferred with two other surgeons (Dr. E.H. Gregory and Dr. P. Tupper), both of whom concurred that the surgical plan seemed reasonable. The patient was also under the care of Dr. J.W. Whipple (orthodontist). Dr. Blair had discussed the feasibility of a surgical solution for mandibular prognathism with Dr. Angle (the patient had also consulted with Dr. Angle before surgery), and Dr. Angle “had advised such an operation.”³⁸

The surgery was carried out at the Missouri Baptist Sanatorium. The surgery commenced with the patient under chloroform anesthesia at 9:20 AM with the patient in the recovery room by 10:30 AM. During the operation, the patient's head was hyperextended over the end of the operating room table. Skin incisions below the inferior border of the mandible on each side provided exposure. The incisions extended up into the mouth and involved cutting through



• **Figure 2-19** **A**, Photograph of Simon Hüllihen (1810-1897). **B**, Illustration of anterior mandibular dento-alveolar segmental osteotomy proposed and **C**, carried out by Dr. Hüllihen. **A** from Ambrecht EC: Hüllihen, the oral surgeon. *Int J Orthod Oral Surg* 23:377, 511, 598, 711, 1937. **B** and **C** from Hüllihen S: Case of elongation of the under jaw and distortion of the face and neck, caused by a burn, successfully treated. *Am J Dent Sci* 9:157, 1849.



• **Figure 2-20** Photograph of Vilray Blair (1871-1955).

the mucous membrane. A full-thickness osteotomy was to be completed on the left side of the mandible between the first and second bicuspid and on the right side between the second bicuspid and the first molar. The osteotomy was initiated with a “double-bladed saw” three quarters of the way through on each side; the inferior border remained intact.³⁸ This allowed for the presence of a stable mandible on each side before the cut was fully completed on either side. A drill was used to place holes on either side of the osteotomy site for eventual “cooper wire fixation”³⁸ near the inferior border on each side before cutting through the inferior border on either side. After the completion of both full-thickness osteotomies, soft gutta-percha was placed over the mandibular dentition, and the upper and lower teeth were firmly wired together through the gutta-percha. This method of intermaxillary fixation immobilized the distal mandible in its new position for the closure of the open bite. Dr. Blair stated that “this method of fixation was suggested to him by seeing Dr. Angle’s fracture bands.”³⁸ The fracture band method of mandible fracture management was commonly in use.

Dr. Blair stated that, as the patient was emerging from the anesthesia, the vomiting of mucus and blood occurred, thereby necessitating the cutting of the intermaxillary fixation.³⁸ After the vomiting subsided, rather than reapplying intermaxillary fixation, a Barton bandage of plaster was used to secure the mandible into occlusion with the maxilla.²¹ When the Barton bandage was removed several weeks later, Dr. Blair felt that there was “bony union on the right side and a very slight but perceptible motion on the left side when the fragments were grasped.”³⁸ Dr. Blair decided to take the patient to the office of the treating dentist (orthodontist), Dr. Whipple. He requested that Dr. Whipple place two dental bands with a bar between them across the weak left osteotomy site. This would afford the patient sufficient support and allow him to go without the additional protection of a Barton bandage or intermaxillary fixation.” Unfortunately, while he was tightening the bands across the left and right osteotomy sites, Dr. Whipple “broke the union of the provisional callus.”³⁸ At this point, Dr. Whipple applied properly adjusted bands to the teeth on the left side of the mandible, and Dr. Blair reapplied a Barton plaster external facial bandage.³⁸ Dr. Whipple also placed crowns on the mandibular teeth posterior to the osteotomy sites so that the molars would occlude properly with the upper teeth, and he filled some points on the teeth to improve the occlusion of the incisors.³⁸ Apparently, the combination of the mandibular setback with the clockwise rotation resulted in “a small [osteotomy] gap between the second bicuspid and first molar on the right side” but “without a gap [previously edentulous space] in the left first bicuspid region.”³⁸ Despite these difficulties, the osteotomies went on to heal with a reasonable correction of the occlusion.³⁸ Dr. Blair stated that there was “almost a complete loss of cutaneous sensation of the lower lip” but that the “sensation of the tooth-pulp is good.”³⁸

Interestingly, Dr. Whipple, the treating dentist and orthodontist, published his version of the patient’s surgery, convalescence, and ultimate result in *Dental Cosmos* in 1897.²¹² This was before the journal publication by Dr. Blair, in which he gave a full account of the case from his perspective.³⁸ Although Dr. Whipple’s description of events varied somewhat from what was eventually reported by Dr. Blair, both concurred that the procedure had taken place and that it was a success.

In his textbook, Dr. Blair cautions the surgeon that “the lower jaw should be set back only far enough to be in harmony with the rest of the face” and that the surgeon should “leave it to the orthodontist to bring forward the upper incisors if necessary.”⁴⁰ He stated that “an orthodontist should be involved in the planning of the patient’s treatment from the beginning.”⁴⁰ For some patients, Dr. Blair notes that “it would be of considerable advantage to have the upper jaw [orthodontically] expanded and the upper incisors and canines brought forward before the lower jaw is surgically set back.”⁴⁰

A horizontal osteotomy through the ascending ramus, on each side and above the occlusal plane, as a method of

“setting back” the prognathic mandible was first described by Lane.¹¹⁷ During the early 1900s, Blair³⁹ and Babcock¹⁹ also made use of the horizontal ramus osteotomy on each side for the correction of prognathism.³⁶

In 1912, Harsha reported bilateral wedge resections of the mandible body near the angle regions to set the mandible back; this was similar to the procedure that Blair described in 1897.⁸⁹ Unfortunately, at least one of his reported patients suffered the loss of (and the eventual need to remove) the entire anterior part of the jaw. In 1945, Moose¹⁴³ may have been the first to describe a fully intraoral technique to carry out horizontal ramus osteotomies for the correction of mandibular prognathism. In 1950, G.V. Barrow and Reed O. Dingman discussed the surgical management of mandibular prognathism.²² They described a method to determine the exact amount and shape of mandibular bone to be resected on each side by using dental models that involved the construction of splints and templates. Their described meticulous approach was similar to that of Angle in the 1903 edition of his textbook.⁷ The operation was reported by Dingman as having a two-stage approach^{71,72}; this was to separate the intraoral and extraoral aspects to maintain control of the segments and to limit the risk of infection. During the first stage, the teeth to be extracted (i.e., the first molars) were removed with the use of local anesthesia. As determined by the “preconstructed template,” osteotomies for wedge resection were initiated through the intraoral approach at the time of the extractions. Approximately 4 weeks later, the patient was anesthetized in the operating room for the second stage. An incision of the skin of the neck was made 2 cm below the inferior border, and the lower aspect of the mandible was exposed on each side. The previous intraoral vertical cuts on each side were identified. With a power-driven bur or saw, the two vertical cuts were continued through the inferior border. Attempts to preserve the inferior alveolar nerve were made. Stabilization across the osteotomy on each side site was with 24-gauge stainless-steel wires. The teeth were placed into occlusion with the use of either orthodontic appliances or Erich arch bars. In either case, to improve accuracy, the mandibular dentition was secured into a prefabricated acrylic wafer before intermaxillary fixation occurred.

In 1954, J.B. Caldwell and G.S. Letterman devised a vertical osteotomy of the ascending ramus that involved the decortication and perforation of the fragments and then the splitting of the medial and lateral cortices of the ramus to allow for setback of the mandible followed by direct wire fixation.^{45,46} This innovative technique had the advantage of providing at least a degree of overlapping of the bone for more reliable and rapid healing.

All of these techniques for the management of mandibular prognathism^{79,87,91-93,96,138,167,211} eventually fell by the wayside after the introduction by Hugo Obwegeser of what we now refer to as the “intraoral sagittal splitting of the mandibular ramus,” which is described later in this chapter.¹⁴⁹ The stabilization of the osteotomy segments is now generally accomplished with titanium bicortical screws

or with a titanium plate and screws in accordance with the pioneering innovations of H. Luhr, who is also discussed in a later section of this chapter.¹²⁷

Mandibular Advancement for Retrognathism

An important early maxillofacial textbook publication was *Surgery and Diseases of the Mouth and Jaws* by Vilray P. Blair (Fig. 2-21).⁴⁰ At the time of publication, Dr. Blair was a Major in charge of the subsection of Plastic and Oral Surgery, Section of Surgery of the Head Office of the Surgeon General of the Army in Washington, DC.¹⁹⁷ He was also a professor of oral surgery at the Washington University Dental School and an associate in surgery at the Washington University School of Medicine in St. Louis, Mo. In his textbook, Chapter 21 deals with the treatment of deformities and malrelations of the jaws.

Dr. Blair cautions that, before entering into jaw-straightening surgery, the surgeon would be well advised to do the following:

- To not undertake any cases without first having the fullest confidence in his or her patient
- To remember that procedures undertaken for only “moderate” deformity should not be taken lightly
- To remember that maximum restoration will also require orthodontics
- To remember that the earlier a competent and genial orthodontist is associated with the case, the better it will be for both the surgeon and the patient

In the 21st century, these remain useful words of wisdom for the surgeon to consider before entering into the treatment of a patient with a jaw deformity.

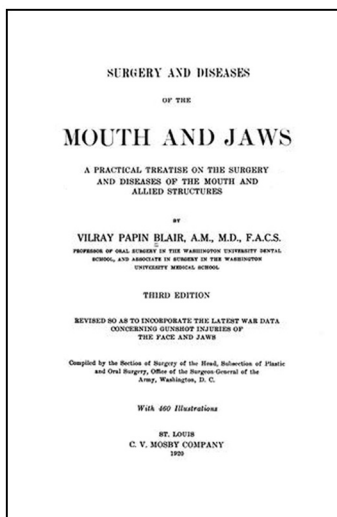
According to Dr. Blair, for the treatment of mandibular micrognathia, a surgical procedure may be used to “bring

forward the lower jaw to harmonious outline” with the maxilla.⁴⁰ Malocclusion in a patient with a jaw deformity results from the “pressure and counter pressure of growth and apposition.”⁴⁰ Normal occlusion cannot be established by jaw surgery alone. To reach certain objectives, the surgeon “shall have knowledge of occlusion and of the scope and limitations of orthodontic procedures.”⁴⁰

Blair went on to say this: “We have to accept the upper jaw position but the lower jaw is capable of almost any kind of surgical adjustment. The complication of an ununited fracture of the lower jaw is rare. The complication of necrosis or loss of teeth from surgically sectioning the ramus is not reported in the world literature.”⁴⁰ However, “osteotomy of the ramus of the mandible is a recognized procedure for the treatment of temporomandibular joint ankylosis.”⁴⁰ Esmarck, who was a prominent surgeon when Dr. Blair was discussing this treatment, recommended the removal of a whole section of the horizontal ramus to avoid a union (i.e., reankylosis) when creating a pseudoarthrosis for the treatment of temporomandibular joint ankylosis. However, Blair felt that surgeons did not need to concern themselves with the possibility of necrosis or nonunion when “completing an osteotomy through the inferior alveolar neurovascular bundle.”⁴⁰

Blair felt that the surgeon could correct mandibular retrognathia by completing a ramus osteotomy on each side and then “bringing the distal mandible forward to meet a good occlusion with the maxillary arch.”⁴⁰ Intermaxillary fixation could then be achieved with stainless steel wires and the use of soft cement in between the occluding teeth.⁴⁰ Fixation across the osteotomy site was felt to be unnecessary. The (horizontal) ramus osteotomy was made at or just above the mandibular occlusal plane with the use of a Gigli saw placed transcutaneously.³⁹ The osteotomy was cut through the inferior alveolar neurovascular bundle, which was not felt to be important either to the circulation of the mandible or as a cause of disability. The ramus osteotomies were later made blindly and transorally with a Gigli saw.⁴⁰

According to Dr. Blair, “the operation presented three distinct problems: (1) the cutting of the bone [which is the easiest of the three]; (2) the placing of the jaw into its new position; and (3) holding it there [for the time required for healing].”⁴⁰ He used postoperative x-rays to show that the fragments of the ramus remained in contact at the posterior border of the osteotomy. “[A]s the distal mandible is brought forward,” said Dr. Blair, “a gap is created anteriorly while the fragments remain in contact at the posterior border. For healing to occur, the bone gap must be filled with granulations. The resulting bone scar tends to contract [relapse] for months afterwards.”⁴⁰ The maintenance of tight intermaxillary fixation was felt to be essential to maintain the new position. Blair used iron wire for its strength, lack of stretch, and pliability; it could also be twisted, unless it was damaged by sharp instruments.⁴⁰ He also used quick-setting cement between the occlusal surfaces of the molars to prevent the placement of postoperative forces on the anterior teeth, which could cause the patient significant pain.⁴⁰



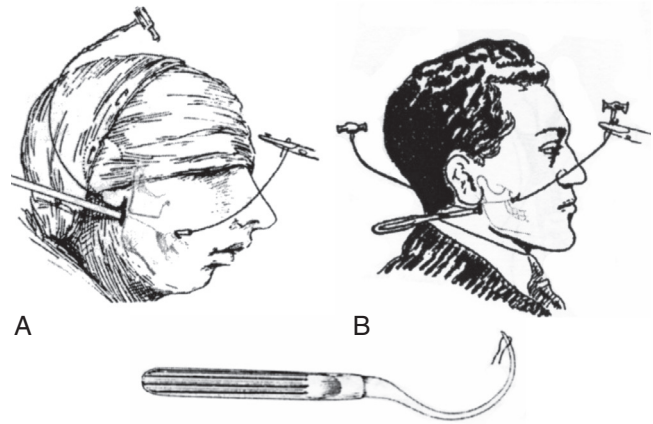
• **Figure 2-21** Vilray P. Blair published a text entitled *Surgery and Diseases of the Mouth and Jaws*, the first edition of which was published by the C.V. Mosby Company in 1912.

Dr. Blair also used this same operation involving horizontal ramus osteotomies for the treatment of ankylosis associated with severe mandibular retrusion; this operation was first carried out by Dr. Mudd with Dr. Angle assisting in 1897. Dr. Blair also suggests addressing the residual chin deformity by either “injecting paraffin into the chin or by inserting a piece of cartilage or rib.”⁴⁰

Most of the early reported attempts at horizontally advancing the retrognathic mandible focused on the mandibular body. Dr. Blair specifically addressed the problem of an anterior open bite by rotating the distal mandible counterclockwise after body osteotomies and ostectomies. He states that the “sectioning of the mandible on both sides in front of the first molars [as long as the molars are in occlusion] is the preferred procedure. The anterior mandible can then be moved into good occlusion with the upper teeth. In other cases, it is necessary to remove a V-shaped section of the bone on each side with extraction of the tooth [bicuspid]. The apex of the V-shaped ostectomy segment is at the lower border of the mandible and usually a tooth is extracted from the site of the section on each side. The bone is cut using a Gigli saw or a cross-cut Fisher bur.”⁴⁰

Dr. Blair goes on to state that “the deformity is in both jaws but the operation is limited to the lower jaw. It is best to restore the lower jaw to its proper form and then manage the residual open bite by bringing down the upper incisors with orthodontic appliances or by extending the upper teeth with porcelain crowns. Fixation is achieved with a prefabricated splint constructed on plaster dental models. This will prevent pulling down of the distal segment by the digastric and geniohyoid muscles.”⁴⁰

A variety of techniques were carried out by pioneering surgeons to address concerns regarding the maintenance of contact between the fragments to achieve bony union; soft-tissue coverage to limit infection; opposing muscle pull to limit relapse; and adequate fixation. In 1928, a “step osteotomy” of the body of the mandible was proposed by Von Eiselberg.²⁰¹ Also in 1928, Gadd reported a stepped procedure for the body of the mandible for the purpose of correcting asymmetric mandibular deficiency.⁸⁰ That same year, Limberg completed L-shaped sliding osteotomies of the mandibular body to improve bone-to-bone contact after advancement.^{122,123} In 1931, Kostecka published his work with the Blair-type transcutaneous horizontal ramus osteotomies with the use of a Gigli saw (Fig. 2-22).¹¹⁵ In 1936, Kazanjian performed mandibular “step” osteotomies anterior to the mental foramen with extension back into the body of the mandible to allow for advancement and fragment approximation (Fig. 2-23, A through D).^{68,103-108} The constant problems encountered with all of these approaches involved maintaining sufficient bone-to-bone contact and achieving adequate fixation for a stable long-term result. The problems were finally solved through the pioneering work of Hugo Obwegeser (i.e., the intra-oral sagittal splitting of the ramus of the mandible) and Hans Luhr (i.e., plate and screw fixation across the osteotomy site), which is described later in this chapter. In 1960, Caldwell described vertical osteotomies of the ramus with interpositional iliac



• **Figure 2-22** As originally depicted, methods for the correction of mandibular deformities by the blind technique. **A**, The method of Blair. From Blair VP: *Surgery and diseases of the mouth and jaws*, ed 3, St. Louis, 1914, C.V. Mosby Company. **B**, The method of Kostecka. From Kostecka F: *Die chirurgische therapie der progeneri. Zahnaerztliche Rundschau* 40:669–687, 1931.

bone graft for the correction of micrognathia.⁴⁵ A C-osteotomy of the ramus was later described by Caldwell and colleagues, in 1968.⁴⁶ Although these osteotomies with interpositional grafting are occasionally used for the management of mandibular retrognathism, the Obwegeser sagittal split ramus osteotomy is generally preferred. Throughout the 18th and 19th centuries, many surgeons saw the need to mobilize the maxilla for better access either to remove an epipharyngeal tumor or for the correction of maxillary deformity.

Maxillary Osteotomies for Management of Dentofacial Deformity

In 1868, David Williams Cheever of Boston published a report of a Le Fort I osteotomy for the purpose of exposing and removing a large nasopharyngeal polyp (Fig. 2-24).^{55,88} After completing the tumor resection, the maxilla was placed back in its original location. Cheever stated, “so far as I know, the operation including both superior maxillary bones—is novel.”⁵⁵ “Nothing but the posterior attachments of the upper maxilla now prevented their depression and hinging on the pterygoid processes. The upper jaw was brought down so as to expose the tumor.”⁵⁵ By the early 1900s, reports of series of patients undergoing maxillary osteotomies for the purpose of tumor resection were detailed in the literature.³⁷

With reference to the treatment of dentofacial deformities, in 1921 in Berlin, Cohn-Stock was the first to report an elective maxillary osteotomy to establish a preferred occlusion.^{56,214} He used a two-stage approach (a precaution against flap necrosis) when completing an anterior maxillary osteotomy. Martin Wassmund, the founder of the “German school” of maxillofacial surgery, is credited with developing the one-stage anterior maxillary osteotomy.²⁰⁵⁻²⁰⁷ The Wassmund procedure was completed through limited palatal and labial incisions with subperiosteal tunneling. The tunneling