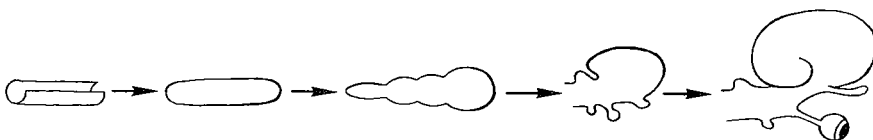


## 57. Median Cleft Lip with Hypotelorism

DEMYER'S group I is characterized by a median cleft of the lip and absence of the premaxillary bone, nasal septum, nasal bones and crista galli. The ethmoid bone, which sets the interorbital distance, is hypoplastic, resulting in orbital hypotelorism. The eyebrows join each other across the midline. Occasionally, the forehead reveals a median keel-like protrusion known as trigonocephaly. The secondary palate may or may not have a median cleft. This group has an unmistakable facies, and it is pathognomonic of a severe congenital brain defect with amentia.

### THIS FACE PREDICTS THE LACK OF BRAIN

DeMyer has a simplified delineation of the central nervous system as a tube sealed off at each end. Normally, the rostral end bulges out like an expanding balloon which undergoes diverticularization into paired cerebral hemispheres, into paired optic bulbs and into olfactory bulbs.



In this anomaly there is serious curtailment of diverticularization. Instead of dividing into hemispheres, the cerebrum remains holospheric like the original simple balloon, so the generic term for this process is *holoprosencephaly*.

The olfactory diverticula are almost always absent in such a brain, a condition which earns the term *arhinencephaly*. The arrest in diverticularization can occur at any stage. When there has been no hemispherization of the brain, the interhemispheric fissure is absent and there are no lobes, then the malformation is *alobar holoprosencephaly*. If the arrest occurs at a later stage and the face and brain are better developed—the interhemispheric fissure extending to the frontal poles, which still keep their continuity across the midline—the malformation is *lobar holoprosencephaly*.

Although median cleft lip with orbital hypotelorism is the commonest facies with holoprosencephaly, it is merely one stage in a spectrum of malformations. In 1967 C. J. Kurlander, W. DeMyer and J. A. Campbell diagramed a fascinating group of facies of holoprosencephaly.

Facies I is the severest degree of orbital hypotelorism, revealing *cyclopia* with a single median eye in a single median orbit. A subgroup showed a single globe in a single orbit but with two corneas.

Facies II is *ethmocephaly*, with the orbits completely separated and the tubed proboscis having migrated from the forehead to between the eyes.

In facies III or *cebocephaly*, the proboscis lies on its side but is still a fleshy tube lacking skeletal support. DeMyer calls the nasal shift from I to III the “march of the proboscis.”

In facies IV, the proboscis has been replaced by nares, but the nasal bones and septum are absent. In facies I through III, although the lip is not cleft, it has no true philtrum and the premaxillary bone is missing. Facies I through IV, according to DeMyer, are almost invariably associated with alobar holoprosencephaly, although one of his cases in IV had lobar holoprosencephaly.

In facies V there appears the intermaxillary segment consisting of rudimentary prolabium and premaxilla which removes it from



I. Cyclopia



II. Ethmocephaly



III. Cebocephaly



IV. With median cleft

the median cleft grouping. Facies V has a variable expression of holoprosencephaly, sometimes arhinencephaly or even a brain that is generally normal but usually small and deficient in developmental potential. DeMyer states that the median cleft lip facies IV is pathognomonic of holoprosencephaly, for among 30 autopsied cases in the literature and 14 of his own there have been no exceptions. Occasionally, holoprosencephaly occurs without a warning facies so that DeMyer editorialized:

The face invariably predicts the brain, but the brain does not invariably predict the face.

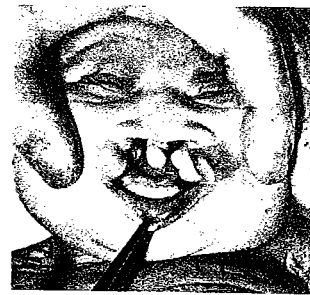
### DIAGNOSTIC CONFIRMATION

Confirmation of orbital hypotelorism is obtained by measurement of interorbital distance on posteroanterior x-ray films of the skull. Electroencephalography will show abnormal patterns. Transillumination is also possible.

### TREATMENT

Treatment of median cleft lip with holoprosencephaly is usually not in the realm of plastic surgery. As pointed out by DeMyer, patients in facies I through III invariably die in the neonatal period, and those in facies IV usually die in the first year. Patients in facies V may survive to adulthood, but death in the early years of childhood is likely. Infants with alobar holoprosencephaly, facies I through IV, are amened, whereas infants with lobar holoprosencephaly, facies V, may show slight developmental progress but will be hopelessly retarded.

Patients with median cleft lip and holoprosencephaly have difficulty with feeding, temperature control and convulsions. DeMyer notes that a regular infant diet can be administered through a bottle or Asepto syringe, that oscillations in temperature need not be treated with antibiotics unless a specific infection is diagnosed and that phenobarbitol or Dilantin need be given only after the onset of clinical seizures.



V. Rudimentary prolabium and premaxilla



## GENETIC COUNSELING

From reports in the literature there is evidence that, once a child with median cleft lip and holoprosencephaly has been born in a family, the risk of another affected child is high. For this reason, DeMyer advises that the parents be warned of this reproductive risk so they can make a rational decision whether to procreate or adopt.

## UTILITARIAN SURGERY



As I noted in 1968, all of these patients are without mentality, do not survive beyond infancy and thus do not require corrective surgery. Once the diagnosis of alobar holoprosencephaly has been made, the prognosis is sealed and the parents should know that only one in a hundred survives the first year. It is far better that the infant be placed in an institution. If the parents insist on keeping the child, and if it survives more than the usual period, closure of the median lip cleft is possible. Extensive facial reconstruction, of course, is not warranted, but any large median gap in the upper lip is ideal for a lower lip-switch flap. When the columella is missing, it can be partially constructed by extending the distal portion of the flap. The lack of nasal support will certainly prevent an aesthetic result, but closure of the cleft does facilitate feeding and improves appearance for the family's relief during the indefinite existence of the patient.



This semilobar holoprosencephaly patient with small brain, hypertelorism, flat nose with absence of septum and columella and median cleft of the lip and alveolus had a primary Abbe flap with a columella extension to facilitate feeding. The patient lived to the age of 18 months and the case was published in *Plastic and Reconstructive Surgery*, July 1968.

C. R. Dehaan, for Stark in 1968, reported on a median cleft of the lip associated with microcephaly, arhinencephaly, orbital hypotelorism, bilateral exophthalmos, absence of the entire central portion of the lip (including the premaxilla but with a normal palate posterior to the incisive foramen) and a single central ostium leading into a nasal cavity devoid of vomer, septum and ethmoid plate. He had treated four "true" median clefts which were actually of the pseudomedian or "false" type. He described the surgical treatment:

The clefts were repaired by a simplified straight-line technique during the first week of life to facilitate feeding. The infants tolerated the surgery well, but none survived longer than 5 weeks.

C. J. T. Pinto and K. S. Goleria of Bombay, India, in 1971 at the Congress in Melbourne added three agenesis cases with median cleft lip and holoprosencephaly to the world records, stating that for these no surgical treatment had been indicated or administered.

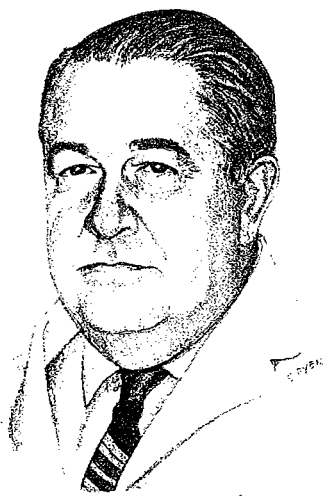
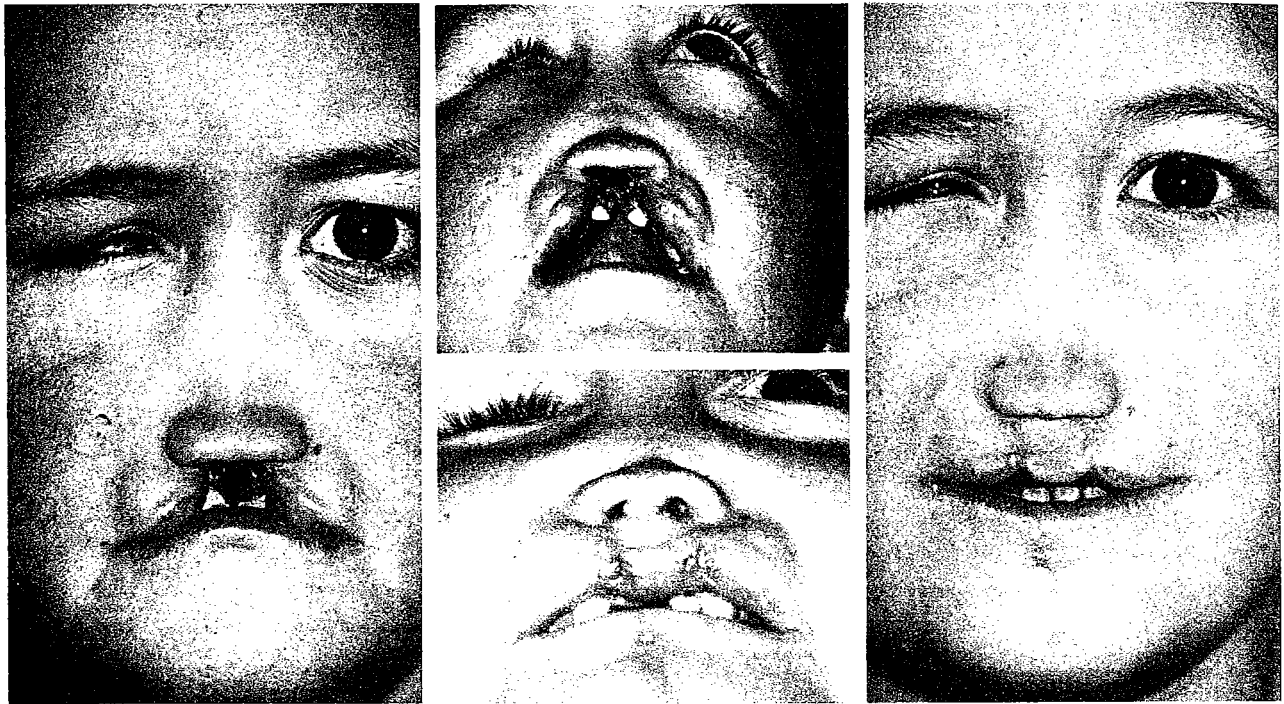
#### FOUR EXCEPTIONS

The cases of B. Vilar-Sancho Altet, Jacob Longacre, Jack Fisher, and James Wells constitute four exceptions. B. Vilar-Sancho Altet of Madrid, Spain, reported a case in *Revista Española Cirugía Plástica* in 1968 that certainly confuses the issue and breaks DeMyer's rule that *the face invariably predicts the brain*. A nine-year-old male had agenesis of the right eye and hypotelorism, elongation of the middle third of the face with absence of the dorsum of the nose as well as absence of the premaxilla and prolabium. The patient also had tetralogy of Fallot but other-



wise was normal, even in intelligence. Vilar-Sancho Altet admitted that had he first seen the patient as a newborn he would have diagnosed his condition as arhinencephaly and postponed surgery.

Treatment: The median cleft of the lip was closed with an Abbe flap, the distal portion being used to construct the columella. Both Vilar-Sancho Altet and I reported the same approach on median clefts in the same year, 1968.



*Jacob Longacre*

The generous Jacob J. Longacre of the University of Cincinnati collected rare books and rare cases over the years until he accumulated the third largest private plastic surgery library in the world. He treated some very fascinating patients. Cognizant of his treasure chest of cases, I requested a median cleft and was rewarded with this rarity treated with split-rib grafts and this comment:

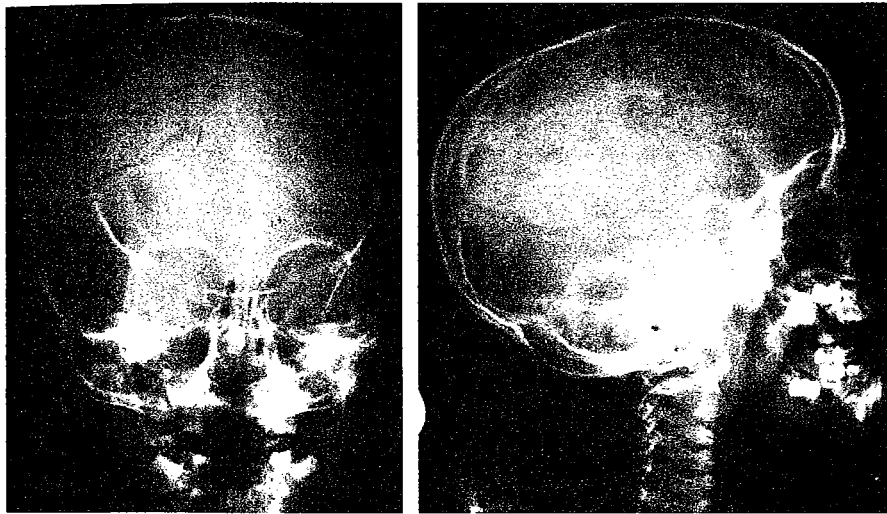
In review of the literature I have failed to pick up another case which following survival was so treated.

As crowned champion of the split-rib graft, Longacre first used the principle in 1952 and applied it to a variety of deformities, publishing about 20 papers on the subject. In December of 1974 he kindly forwarded this unpublished case report.

This child was born with a complete midline defect with absence of premaxilla, prolabium and columella with associated cleft lip and cleft palate deformity. The findings of midline cleft, hypotelorism, and microcephaly are most compatible with a diagnosis of arhinencephaly. The lip was repaired at five months and revision of the lip repair by Dr. O'Malley in Orlando at age one year. Motor and sensory development was slow; the patient was still crawling when first seen at age two. Frontal and lateral x-rays revealed the microcephalic skull with the grossly deformed and hypoplastic maxillae and malars with associated midline cleft and absence of the premaxilla. The interorbital distance measured 15 mm. due to hypoplasia of the ethmoids.



5 months



2 years

Following expansion and cleft palate repair by Dr. Longacre at age four, he developed a vocabulary of 250 words within six months and two of the words he used constantly were "no nose, no nose." A columella was reconstructed with local tissues and the defect of the middle third of the facial skeleton reconstructed in two stages with split-rib grafts to malar, maxillary and nasal regions improving patient's appearance at age four and one-half, and six.

Bilateral ptosis of eyelids still gave the patient a very stupid appearance. This was corrected with fascial slings tied into the frontalis muscle during the operation for insertion of the third set of onlay split-rib grafts. Following this, the patient showed a tremendous spurt of improvement in vocational, academic, and social adjustment. He is now ten years of age and attending regular school. Frontal and lateral x-rays reveal the autogenous bone support provided to the extensive midline defect of the facial skeleton by the staged split-rib grafts.

F.H. Paternal great-uncle and nephew born with unilateral clefts of primary and secondary palate.

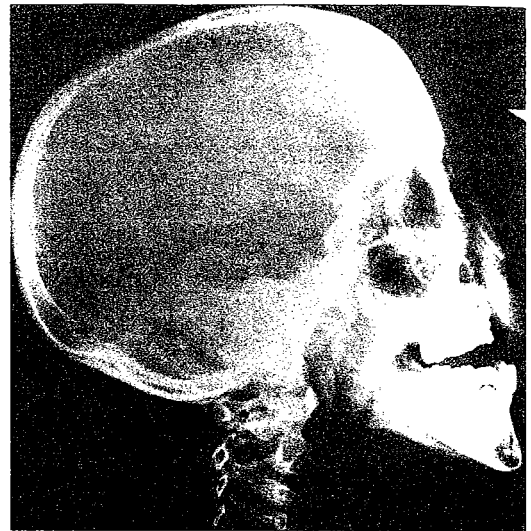
F.T. "Flu"



7 years



10 years



10 years

*An odd coincidence*

During the 1974 American Board of Plastic Surgery examinations in Portland, Oregon, one of my questions directed at Jack D. Fisher was on the subject of holoprosencephaly. Five months later, he repaid me by reporting that he now had a case of a related nature. Subsequently, photographs and the accompanying history and physical findings were forwarded from the University of Virginia Medical Center, Charlottesville, by resident James H. Wells, who with Jack Fisher will be publishing this case report.



This three-month-old female, born of unrelated parents but with a paternal aunt having unilateral cleft lip and palate, was the product of a pregnancy complicated with polyhydramnios and a question of aspirin overdosage at two to three months' gestation. Her birth weight was 5 pounds 5 ounces, her Apgar score 8 at one minute and 9 at five minutes.



Her deformities include a hypotelorism, cleft lip and palate, clinodactyly, and a microcephaly; head circumference was 32 cm. at two months. She had a neuroencephalogram performed, and this is a summary of the radiologist's report.

Impression: This examination shows evidence of severe focal superficial atrophy with an almost porencephalic area in the right parietal region. An absence of the septum pellucidum, slightly fuller ventricles than one would expect in this age, and midline cerebellar agenesis or atrophy.

It was reported by Wells:

To date, she appears to have a normal growth and development pattern. She is eating well, gaining weight, utilizing a Lamb's nipple.

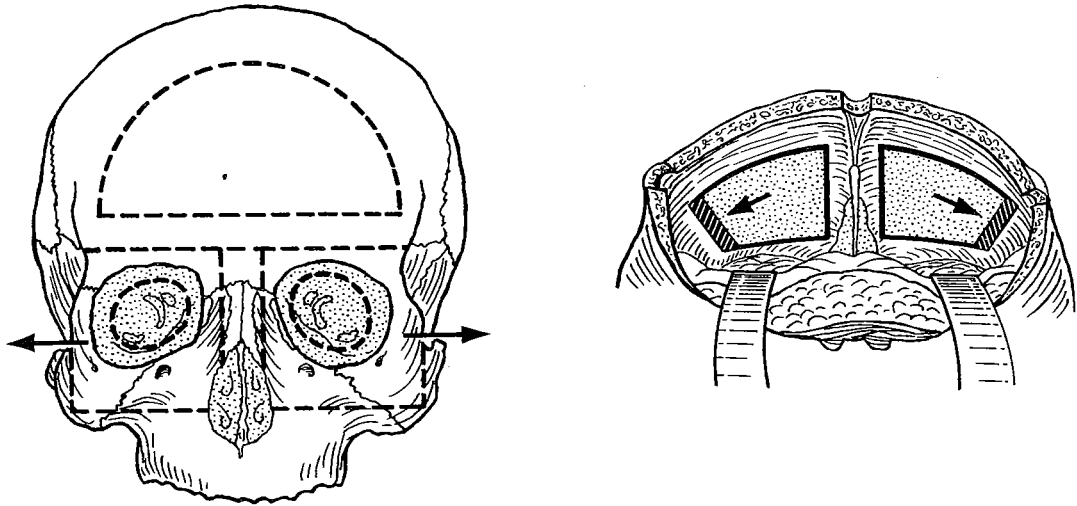
Discussion with Fisher indicated his plan of bilateral rotation of the lateral lip elements with closure of the muscles in the midline. This seems a logical first step, and if the patient shows reasonable progress, then an Abbe flap with an extension could create a philtrum and columella. Craniofacial surgery in a reverse Tessier and split-rib grafts might be indicated in this case and possibly in that of Vilar-Sancho Altet.

#### SURGICAL TREATMENT OF HYPOTELORISM

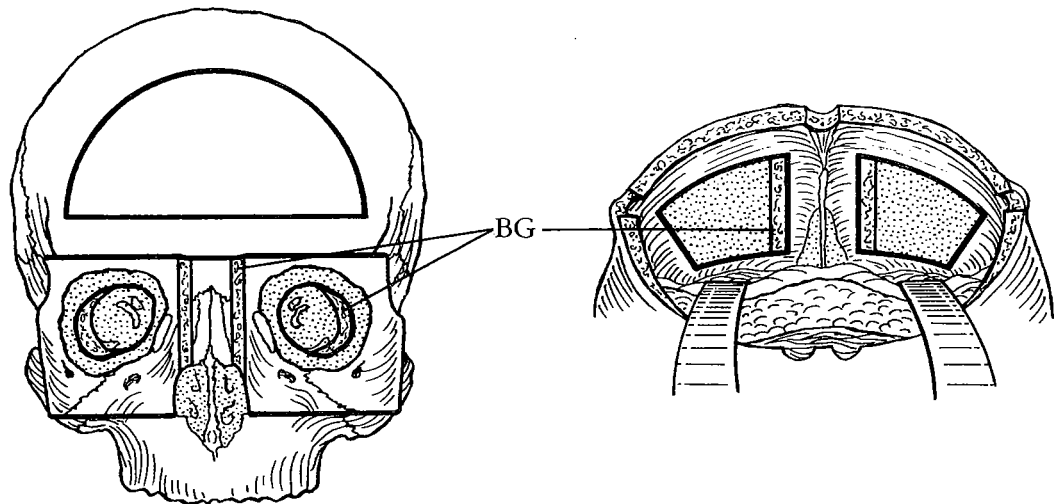
Although most cases of orbital hypotelorism will not deserve surgical correction, as noted there are exceptions. In 1975 in *Plastic and Reconstructive Surgery*, Converse, McCarthy and Wood-Smith reported the first correction of an orbital hypotelorism. Although no cleft was involved, the case and the treatment are of interest.

The patient was a Caucasian girl born with orbital hypotelorism and hypoplasia of the nasomaxillary region. The nose was flattened, the columella shortened and the septum absent. She received grafts of autogenous cartilage, composite earlobe and preserved cartilage to her nose between the ages of two and nine years. At 14 years she revealed an intercanthal distance of 19 mm. and radiographic interorbital distance of 14 mm. (24 mm. expected for this age). A diagnosis of nasomaxillary dysostosis (Binder) was made.

In 1972 surgical correction was undertaken through a combined craniofacial route, similar to that for correction of orbital hypertelorism. The bony orbits were mobilized by superior horizontal osteotomies, vertical, inferior horizontal and paramedian osteotomies and a transverse cut of the orbital roof. Segments of bone were removed from the lateral aspect of each roof.



The orbits were then translocated laterally for a total distance of 8 mm. The resulting defects in the roof and in the nasofrontal areas were filled with iliac bone grafts [BG].



The patient subsequently had an inlay nasomaxillary skin graft, and the total surgery resulted in a definite improvement in her appearance.