25. *Genetic Counseling, Parent Guidance, and Preoperative Preparation*

WHEN an infant with a cleft of the lip and palate appears in the newborn nursery, a chain of reactions and actions is set in motion. After nine months of happy anticipation the parents listen in horror to the news that their baby has a facial deformity. If either parent had suffered the deformity or knew of it in the family, then the gnawing fear already present has become a painful reality. If the anomaly is unknown to them, it will strike panic in their hearts. The disappointed obstetrician will call in the pediatrician, and he, in turn, will call a plastic surgeon.

The future of this baby now becomes the responsibility of the plastic surgeon, who will be assisted, as the baby develops, by the prosthodontist, orthodontist, pedodontist, otolaryngologist, speech therapist and even the psychiatrist.

GENETIC COUNSELING

It is well to put the parents somewhat at ease by assuring them that in no way, by negligence or other obvious action, have they as parents been responsible for their baby's anomaly. It should be explained that the ratio of cleft to normal births runs about 1:750 in the United States and that, although there is a definite hereditary factor, it is only a part of the entire problem, for parents with one or more children with clefts can have a normal infant and, to a lesser extent, normal parents with no family history of anomalies can produce a child with a cleft. The arrival in a family of one infant with a cleft does not indicate that subsequent babies will be deformed. Although a repetition can occur, in general the chances are against it.

Heredity plays a significant role in the occurrence of cleft lip and palate. CL, with or without CP, appears to be transmitted by separate genetics from that involving isolated CP. Although some data exist suggesting sex-modified inheritance in clefts, they remain inconclusive. The data available support a polygenic inheritance for both CL, with or without CP, and isolated CP. An exception is the rare Van Der Woude or "lip pit" syndrome, in which CL and/or CP occurs with a lower lip fistula and which is transmitted by an autosomal dominant gene.

Woolf, Woolf and Broadbent (1963) in Utah, and Bixler, Fogh-Andersen and Conneally (1971) in Denmark published data from select populations from which it is possible to derive empirical risk factors for concerned parents. The child of a parent having a cleft lip, with or without associated cleft palate, has a 3 to 4 percent risk of being similarly affected. A child with unaffected parents but with an affected sibling has a 4 to 5 percent chance of being affected. The child with both an affected parent and an affected sibling has a 13 percent chance of being affected. The risk for children with normal parents but with an affected grandparent, aunt, uncle or first cousin is under 1 percent.

The analogous data for isolated cleft palate are as follows: Children of an affected parent have a 6 to 9 percent risk, children with normal parents but with an affected sibling have a 2 percent risk, children with both an affected parent and an affected sibling have a 13 to 14 percent risk and children with normal parents but with an affected grandparent, aunt, uncle or first cousin have under a 0.5 percent risk.

An exception to the above data is the "lip pit" syndrome. Seventy percent of those affected will also have CL(P) or CP and will pass on the gene to 50 percent of their children. Of the 50 percent, 60 to 80 percent will have CL(P) or CP.

The Fraser parental genetic guide

The studies of Canadian geneticist Clarke Fraser has made it possible to advise parents of their percentages.

If they are unaffected and have an affected child, the chances

of the next child being affected is 4% in CL ± CP but 2 to 7% in CP depending on the existence of affected relatives. If they as unaffected parents have two affected children, then the chance of the next being affected increases to 9% in CL ± CP and drops to 1% in CP. If one parent is affected and they have no affected children, the chance of the next being affected is 4% in CL ± CP and 6% in CP. If one parent is affected and they have an affected child, the chances of the next baby being affected are 17% in CL ± CP and 15% in CP and if both parents are affected the chance is about 60%. The presence of affected distant relatives apparently does not increase the chance; other major malformations not part of a genetic syndrome reduce the recurrence of the cleft anomaly. The severity of the degree of the cleft increases the recurrence risk by double. For instance, a unilateral cleft lip recurrence risk is 2.5% while a bilateral cleft lip and palate rises to 5.7%.

ETIOLOGY

Extensive study has been under way for years in search of the etiology of this deformity. Both heredity and environmental factors have been covered in the parents' questionnaire.

1. Careful study of the family history for other congenital anomalies.
2. Data on the condition of the parents at the time of conception of the deformed child.
3. Details of the first trimester of pregnancy including the mother's diet and activities, exposure to radiation, occurrence of bacterial or viral infections, use of drugs and experience of emotional stress.

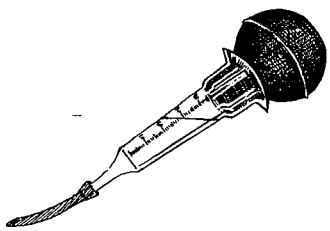
Environmental factors produce clefts in experimental animals, but their influence in man is unproved.

REASSURANCE OF PARENTS

The nurses taking care of the mother and the infant with the cleft will have a natural compassion, but it should be channeled beyond sympathy toward a constructive preparation for what

lies ahead. Poignant memories of other children with clefts who ended up with scarred and twisted lips, crooked hooked noses and cleft palate speech rendered unintelligible by the nasal escape of air can be minimized. The nurse can emphasize to the parents the great progress that has been achieved in the surgical, dental and overall care of these babies.

CARE OF THE BABY



The baby himself can be treated with the same gentle kindness that other more normal babies receive. Only the feeding requires specialized training. A cleft in the lip reduces the baby's ability to suck and will render breast feeding improbable and bottle feeding difficult. A cleft of the palate also interrupts suction and allows escape of fluids through the nose. Therefore, the baby is supported in the nurse's arm with his head held upright in her left hand. Formula is fed by bulb compression through a 50 cc. Asepto syringe with a 1½ inch rubber catheter extension. The catheter is slipped over the baby's tongue, and the formula is fed as the baby is able to take it. As soon as the baby has adjusted to this routine, the mother is instructed in the technique. Once mother and baby are prepared by the nurse, they are allowed to go home. This general routine was taught to me as an intern by MacCollum at Boston Children's Hospital.

In certain particularly malnourished cases in which successful intake seems to be more difficult, a nasogastric tube can be introduced and fixed to the face with tape to facilitate forced and controlled nourishment through gavage.

ANTICIPATION OF SURGERY

Most parents are anxious to have plastic surgery immediately. Yet, when it is explained to them that the best result for their baby can be achieved if the lip and nose surgery is postponed until the tissues are larger and stronger, they are content to wait, take the baby home and fatten him up. If the cleft is incomplete, a definitive closure is scheduled at age three to six months, while a complete cleft is set for an adhesion at three weeks and a definitive closure at six to eight months. It is felt that there

is a better psychological adjustment with fuller understanding of the problem if the parents have a chance to become familiar with the deformity. To shield them completely reduces the maturity of their approach.

SPECIAL CARE BEFORE LIP SURGERY

If the cleft is incomplete, the baby should be at least three months of age, healthy and gaining weight and should have an estimated hemoglobin of at least 10 gm. The requirements can be outlined in a general "rule of over 10":

Weight—over 10 pounds
Hemoglobin—over 10 grams
White count—not over 10,000
Age—over 10 weeks

He should have been trained to sleep on his back in preparation for the postoperative period, when this position is mandatory to protect the healing lip from brushing the bed.

No special preoperative or postoperative formula is required. What was considered satisfactory by the pediatrician before closure will suffice after surgery.

Before we turn our attention to the actual technical aspects of incomplete cleft lip surgery, let us have one last look at another vital principle in craftsmanship.