CLINICAL CONTRIBUTION

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Arhinia

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INTRODUCTION

Among the various types of congenital malformations of the nose, complete absence of the nose is rare (Lütolf, 1976). Rosen (1963) called the absence of the nose alone "arhinia", and the absence of the nose along with complete absence of the olfactory system "total arhinia". Dekaban (1959) and Kemble (1973) described it as often associated with maternal diabetes, hypertension and toxemia of pregnancy. There are various degrees of anomalies in the nasal area which are widely reported in the literature (Coats, 1961; Patten, 1971); but the number of cases of arhinia hardly exceeds ten reports in the English literature in the present century. It is, therefore, worthwhile to study every case. A case of arhinia, bearing relatively few other malformations, is hereby presented.

CASE REPORT

A case of arhinia with absence of the olfactory nerves is described. As far as we know, this is the first attempt at surgical correction at such an early age.

A first-born male newborn, the son of first-degree cousins, was referred to the emergency ward a day after delivery because of respiratory distress (Figure 1). The

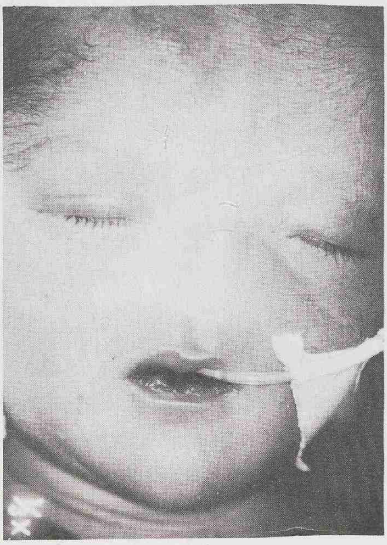

Figure 1. The child on admission. Note hypertelorism, small eyes and the miniature nasal pyramid.

family history did not reveal diabetes, hypertension, congenital anomalies, or any other major illness. The pregnancy was uneventful. Crying after delivery was spontaneous. On examination, the newborn was found to be viable, but breathing was laborious and was accompanied by mild thoracic retractions. Birth weight was 2200 g, head circumference 30.9 cm, chest circumference 28.5 cm, total length 46 cm. Hypertelorism and low-set ears were found. A flat area replaced the nose, without nares, but with a remnant of nasal pyramid which was bony on palpation. The palate was high arched. There was no other oral pathology. Umbilical hernia, hypospadias and pilonidal dimple were the only other pathological findings on physical examination. On neurological examination, a weak highpitched cry and absence of sucking and Moro reflexes were found. Blood and urine examinations were within normal limits. Radiologic examination of the skull, skeletal bones and I.V.P. were normal. Radiologic examination of the nasal area revealed small sized nasal bones and obstruction of the nasal cavity by a large irregular bony mass, located mainly in the posterior part, extending from the ethmoidal to the palatal area, filling the whole width of the nose and fusing with the hard palate (Figures 2, 3). The rest of the nasal area was filled with soft tissue. The neighbouring structures and the inner ear appeared normal. The teeth, as seen in tomographies of the skull, were present and in normal stage of development fitting his age.

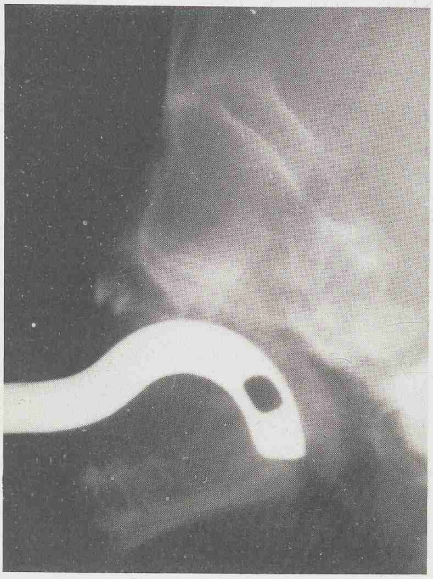

Figure 2. A lateral radiograph. The upper limit of the airway marks the palate. Above the palate a bony mass is filling the whole nasal cavity. A narrow nasopharyngeal cavity is also seen.

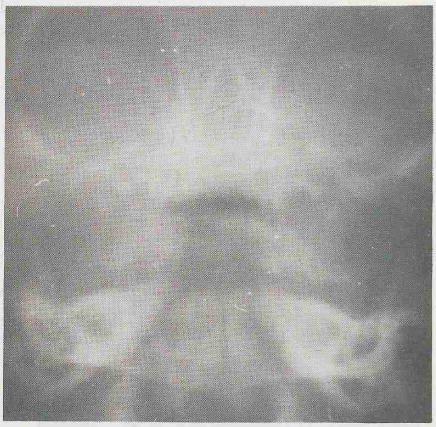

Figure 3. An A-P radiograph. No nasal cavity and maxillary sinuses exist.

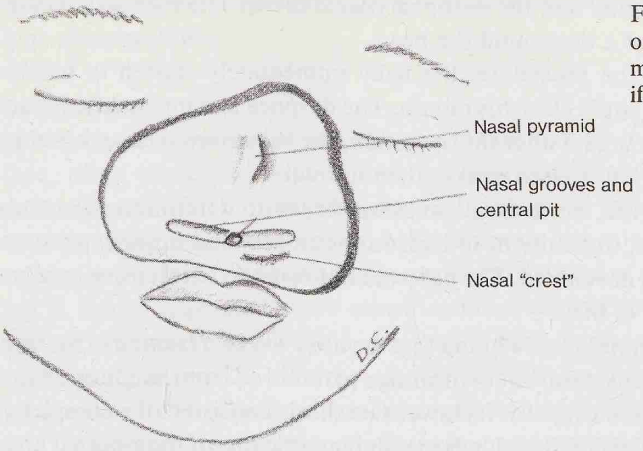

Figure 4. A combination of surgical and post mortem findings, drawn as if the skin was removed.

Electroencephalography, visual evoked response and electroretinography were normal. Feeding could only be effected via an oro-gastric tube, while mild dyspnea persisted. No oral airway tube was needed.

A double-stage surgical procedure was planned: The first stage was to provide an airway and enable the baby to suck. The second was to reconstruct the nose by several procedures. We started the first stage on the 16th day of life. It was decided to try to do the operation under local anaesthesia because it was considered safer due to the patient's general condition. The operation was successfully completed under this anaesthesia. A round incision was made in the area corresponding to the nares. A bony wall was found, corresponding embryologically to the area of the undeveloped septum and cavities, with a small central pit at the site of the nasal spine.

No nasal cavity could be identified and the anterior surface of the bone in between the maxillae was completely flat. The details that could be identified appear in Figure 4. Using bone currets, strong sharp needles and bluntended probes, a passage was created through this bony wall. We tried first to pass through the median pit and then through the bony wall laterally to it in both sides. We faced great difficulty doing this, because instead of the nasal cavity, we were confronted by a dense bony mass which made the advancement impossible. Nevertheless, a narrow tunnel was made in the direction of the nasopharynx, with great difficulty, while monitoring the procedure on television-monitored radiography. It was found that the anterior quarter of the "nasal cavity" was blocked by an unorganized structure of bone, cartilage and soft tissue. The posterior part was completely blocked by a very dense irregular bone which could not be penetrated. A wide polyethylene tube was introduced into the surgically-made tunnel, its posterior half passing submucously between the palate and the "vomer". Posterior rhinoscopy revealed complete blockage of the choanae; but the bony mass

did not extend posteriorly into the nasopharyngeal cavity. The tube was secured anteriorly by means of a tie around the head.

After completion of the procedure, the baby immediately started to breathe through the tube and could close his mouth. The dyspnea and intercostal retractions disappeared. A few days after the operation, the baby started to suck, but his sucking and sucking reflex were weaker than normal.

In the third week of life, his general condition began to deteriorate. Diarrhea, electrolyte imbalance, impairment of liver funtions, bilateral bronchopneumonia, and finally sepsis developed. The baby did not respond to intensive care and died on the 29th day of life.

On post mortem examination, we found that the nose was represented in its upper third by two rudimentary nasal bones forming a pyramid of 3 mm height and 5 mm width, which were fused caudally. Its lower two-thirds consisted of a completely flat bone without any depression. Underneath the remnants of the external nose, the nasal cavity in its anterior part contained soft tissue to a depth of a few millimeters, while all the rest of the nasal cavity was blocked by an irregular hard bone extending from the base of the skull to the palate. The surgically prepared canal for the nasopharyngeal tube passed underneath the hard palate mucosa and then above the soft palate to the nasopharynx. The paranasal sinuses could not be found. The tissue filling the nose and the maxillary sinuses was not histologically examined. Inspection of the brain showed complete absence of the olfactory nerves and bulbs (Figure 5). A thorough study by numerous sections revealed no other abnormality of the brain and the rhinencephalon.

On chromosomal examination prior to death, an inversion in chromosome 9 was

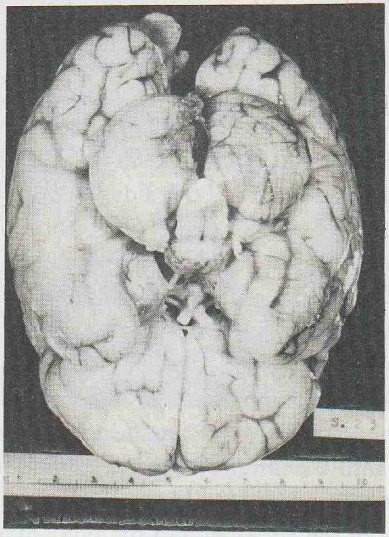

Figure 5. Inferior view of the brain. The olfactory bulbs and nerves are the only missing structures.

found. This chromosomal anomaly is not yet known to be associated with phenotype abnormalities.

DISCUSSION

Arhinia is one of various presentations of malformations in the middle third of the face, often associated with central nervous system anomalies. Probst (1979) summarized different methods, previously suggested by several authors, for the classification of those anomalies. Arhinia is only one minor group in this multifarious group. It may be classified according to the kind of anomalies accompanying it. Probst suggested a new classification system of those anomalies. In this system arhinencephaly is divided into: 1. Pure forms, without accompanying malformations; and 2. Forms associated with malformations. In both subgroups the olfactory bulbs and tracts are absent by definition, the corpus callosum and the fornices are present in the first group and present or absent in the second. No additional facial abnormalities are usually found, except occasionally for clet lip and palate. Our case fits into the first group, although it is not quite clear whether Probst included somatic anomalies in the first or the second group of his classification. However, it seems that this classification method, based on a small number of such cases, may undergo changes when additional cases will be reported.

As to the case presented above, it seems to be a very "mild" case when compared to those previously reported. Many of the arhinia cases suffered from severe central nervous anomalies, sometimes incompatible with life. In the English literature there are only few detailed cases. Post mortems have been performed on only three of them (Marburg, 1943; Dekaban, 1959; Gitlin, 1960). In all three, major brain anomalies were reported. Surprisingly, our case had no macroscopic brain anomalies at all, except for the lack of olfactory nerves and bulbs. As already known, the failure of the development of the nose, olfactory nerves and bulbs is a consequence of disturbance of the invagination of the nasal placodes and olfactory buds. Therefore, there is a direct linkage between those errors (Probst, 1979). The severity of the other malformations in our case were also mild: lowset ears and hypertelorism, high-arched palate, umbilical hernia, hypospadias and pilonidal dimple. Some weakness has been found in the neurological examination. However, the child did survive and died from sepsis. We could not know if death was a result of low resistance or simply due to sepsis.

The surgery done on his nose was for the creation of an airway in order to enable sucking. It is the youngest case in which surgical correction has been performed. The genetic workup revealed no explanation to this anomaly, although recently a report appeared on a mosaic trisomy 9 syndrome with an arhinia case (Kaminker et al., 1985). There is no genetic explanation for a connection between chromosome 9 and a phenotype anomaly.

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