TREACHER COLLINS SYNDROME: Medical and Dental Management: A clinical report

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TABLE OF CONTENTS

ABSTRACT

Treacher Collins Syndrome is a rare congenital craniofacial anomaly affecting 1/50,000 births. It follows an autosomal dominant pattern of inheritance, but a significant number of cases are de novo. Clinical characteristics can vary greatly from one affected individual to another. Malformations mainly involve the parts of the craniofacial complex, but this syndrome is well known for its mandibular hypoplasia. A case is presented, demonstrating the complex problems associated with Treacher Collins Syndrome.

INTRODUCTION

The majority of patients that are seen in a dental practice are healthy individuals. Occasionally, a medically compromised patient will present to the office with complications which the dentist may not be prepared to handle. A child presenting with such problems will often seek treatment from a pediatric dentist who is trained in seeing children with special health care needs. One of the more complicated and seldom seen problems is the patient with craniofacial anomalies. These children usually possess multiple medical problems and thus can be quite challenging to the pediatric dentist. Cases such as these are rare, and are often not reported in the literature. The following is a report of a case involving a patient with Treacher Collins Syndrome. A history of the disease will be presented with the case and discussion focused on the medical and dental management of the patient.

REVIEW OF THE LITERATURE

History

Treacher Collins Syndrome is characterized by multiple developmental abnormalities of the

craniofacial complex. It was first described by Thomson[1] in 1846 and again reported in 1888 by Berry[2]. The major credit for discovery of the disease is given to Treacher Collins[3], who described the main features of the syndrome in 1900. In 1944 and 1949, Franceschetti and co-workers[4] gave this syndrome the name mandibulo-facial dysostosis, after publishing extensive reviews of several family histories. In his report, he described several forms of the syndrome: complete, incomplete, abortive, unilateral and atypical.

Etiology

Treacher Collins Syndrome is an autosomal dominant genetic disorder. It is considered rare, affecting 1/50,000 births. It is believed there is complete penetrance and variable expressivity of the gene[5]. Several reports have documented family pedigrees showing the autosomal dominant pattern expressed through generations. Other reported cases have shown affected individuals with no positive family history. It is presumed that approximately 60% of cases are new mutations. Investigation of Treacher Collins Syndrome has indicated an increase in affected offspring from affected females, and a decrease in affected offspring from affected males[6].

Pathogenesis of Treacher Collins Syndrome is related to excessive and/or premature amounts of cell death in those cell populations derived from the first and

second arch ectodermal placodes[7]. Deficiencies of the first and second branchial arches are caused by localized tissue damage and also from inadequate promotion of growth and cytodifferentiation. This occurs during the fourth week of development[7].

Many attempts have been made to locate the gene which causes Treacher Collins Syndrome. However, at this date, it is still not known.

Systemic features

Individuals affected by Treacher Collins Syndrome exhibit distinct characteristic facies. Most obvious is the palpebral fissure sloping downward and forward, depressed cheekbones, deformed pinna, receded chin and distinct downturned mouth with a fish-like appearance. One out of four people affected will have an abnormal tongue-shaped growth of hair which extends to the cheeks[8].

Studies of the calvarium reveal normal development. However, some individuals may exhibit increased digital markings in the presence of a normal suture relationship. The supraorbital ridges may also be poorly developed. The body of the malar bone is usually underdeveloped with no fusion of the zygomatic arches. The malar bone may sometimes be totally absent. Defects in mastoid bone and paranasal sinuses may also be seen.[9,10,11].

A distinct feature of Treacher Collins Syndrome is at the orbital area where you see a severe antimongoloid slant and deficiency of soft tissue and underlying bone in the inferior lateral orbits. This gives the individual a sad, droopy looking appearance. A high number will show coloboma in the outer third of the lower lid. Vision is usually normal[12]. Recently, Wang and co-workers[13] reported seven significant ocular findings after examining 14 affected patients. They include a subnormal horizontal palpebral fissure length and inferomedial displacement of the lateral canthus in primary gaze; further medial displacement (>4.0 mm.) of the lateral canthus with resultant shortening of the horizontal fissure length on forced eyelid closure; partial thickness eyelid colobomata localized in the nasal one half to two thirds of the lower eyelids; bilateral absence of the inferior lacrimal puncta; bilateral blepharoptosis; inferior placement of the palpebral fissure; and, regular astigmatism without any consistent orientation of the axis of astigmatism relative to the lower eyelid defects, blepharoptosis or lateral canthus. Wang proposed that these findings may be helpful in making a definitive diagnosis of Treacher Collins Syndrome.

Auditory problems are quite common in patients with Treacher Collins Syndrome. Deformities occur at both the external and middle ear. The external appearance may vary from mild to severe. In its severe from, the pinna will be deformed, crumpled forward or

displaced. One may also find extra ear tags and blind fistulas anywhere between the tragus and the angle of the mouth[14]. Internally, poor development of the incus and malleus may lead to conductive, bilateral hearing loss between 50-70 dB[15].

The appearance of the nose often may be large with a narrow nares and bulbous tip. Choanal atresia occurs quite often with varying degrees of severity. There is hypoplasia of the nasopharynx, oropharynx, and hypopharynx with a narrow trachea and larynx. This may cause respiratory and feeding problems and complications may lead to death of newborns and infants[8,12].

Mandibular hypoplasia is the most classic oro-facial feature found in Treacher Collins Syndrome. Most often, the undersurface of the body of the mandible will be concave. Failure of fusion of the maxillary and mandibular process, appearing as macrostomia, is sometimes found. It will show up unilaterally or bilaterally. Poor development of the maxilla leads to a high and narrow palate formation with 28% having clefting of the lip and palate[5,16]. This will result in severe dental malocclusions. The malocclusions will vary from widely separated teeth, to hypoplastic, displaced teeth with an open bite. The mouth is usually large but microstomia will sometimes occur.

Skeletal abnormalities not involving the craniofacial complex is sometimes seen with Treacher Collins Syndrome. These include abnormalities of the

radius, ulna, metacarpals, phalanges and spine[14]. Congenital malformations of the heart and G.I. tract have been reported[17].

The mental status of affected individuals is quite good. Mental retardation is usually not common. They are expected to function within the range of normal developement[8,9]. However, deafness, lack of speech and visual problems may delay cognitive, as well as social skills.

Special anesthetic considerations

There have been several reported cases of sleep apnea associated with Treacher Collins Syndrome[18,19]. The narrowing of the airway is commonly seen and is due to pharyngeal hypoplasia. This leads to loud nocturnal snoring, frequent sleep arousal and often times respiratory depression to obstruction. Surgical correction may sometimes improve this condition. Anesthesiologists will usually provide special care when treating these patients.

Differential Diagnosis

It is very difficult to misdiagnose Treacher Collins Syndrome, due to its classic signs and features. However, there are a few anomalies which share some of the clinical features of mandibulofacial dysostosis. These

include Nager acrofacial dysostosis, Wildervanck-Smith syndrome and hemifacial microsomia.

CASE REPORT

Medical Summary

The patient S.J. was born on 9-18-81 at Good Samaritan Hospital in Portland, Oregon. She was delivered six weeks premature to a 17 year old white female. Her mother reported that the pregnancy was uncomplicated until three days prior to birth when she experienced a leakage of amniotic fluid and was admitted to the hospital. She was born three days later through vaginal delivery. Soon after birth, S.J. experienced difficulty breathing and, at four hours after delivery, was transported to Doernbecher Children's Hospital, Oregon Health Sciences University (OHSU), Portland, Oregon. Upon arrival, respiratory distress continued and she was quickly intubated and an I.V. line for fluids was placed. Chest x-rays revealed a right pneumothorax. At this time, she was in critical but stable condition, breathing with assistance from a respirator.

Physical examination revealed the following findings:

Weight: 2.2 kg.
Length: 51 cm.
Head Circumference: 41 cm.
Resp. rate: 40
Heart rate: 150

Cranium: Small fontanelles

Cheeks and Face: Skin tags on the right cheek and two tags anterior to the right ear. Dimple with increase hair anterior to left cheek.

Eyes: Puffy red reflex, slanting of the eyes.

Ears: Low set, pinna well formed.

Nose: Flat bridge.

Midface: Mandibular hypoplasia

Mouth: Small

Oral Cavity: High palate but intact.

Neck: no masses

Hands and feet: Within normal limits (WNL)

Lungs: Good air movement, no rales.

Cardiovascular: No murmur, pulse 2+.

Abdomen: Soft nontender liver, decrease one cm. No splenomegaly.

Genitourinary: Prominent labia minora.

Neurology: WNL, good reflexes.

Extremities were well perfused except for acrocyanosis.

Two days later, after chest x-rays revealed that the pneumothorax was expanded, the chest tube was removed and an oral airway was placed. On 9-21-81, with the patient now in stable condition, an ENT examination was performed. Results revealed choanal atresia (bilateral obstruction of the nares) along with previous notation of skin tags and hypoplastic mandible. The ear canals were very small. The palate was long and high arching. Further cardiology testing revealed a small ventricular septal defect. There were no murmurs

and no signs of congenital heart disease. There were no plans to surgically correct the VSD at this time.

On 9-24-81, after the diagnosis of choanal atresia was made, a surgical procedure to repair the bilateral obstructed airway was performed under general anesthesia. It was noted by anesthesiology, that intubation of the patient was difficult due to her small oropharyngeal space. During the surgery, silastic stents were placed to keep the airway open through the nares. It was anticipated that she would wear the stents for one year. After the procedure, the chest tube remained to assist in breathing. Three days later, the chest tube was removed after establishing that the patient was breathing well through the stents.

The patient continued to be followed in the NICU (OHSU) and at this time, her weight decreased to 1.5 kg. However, her breathing was steadily improving. On 10-1-81, in the NICU, skin tags from the right cheek and the right preauricular area were removed using local anesthesia. The patient tolerated the procedure very well as it was done at bedside.

As the patient continued to be monitored in the NICU, further specialty consults were ordered. Ophthalmology examination again noted short lid fissures, with a mongoloid slant. The patient however was not following movements well from her left eye. A diagnosis of left optic nerve hypoplasia was given. It was not sure at this time whether there was any blindness.

A specialist from Medical Genetics, OHSU, also consulted and an initial diagnosis of Treacher Collins Syndrome was given. Their conclusion was based solely on clinical findings. Chromosome analysis was ordered.

As the patient was followed in the NICU, her condition continued to improve steadily. Cardiology noted that the small VSD remained, but it was hoped that it would close on it's own.

On 10-29-81, it was found that the nasal stents had migrated from there original placement. A short procedure under general anesthesia was performed to replace the stents. The patient tolerated this procedure well. Soon after this procedure and approximately six weeks post delivery, the patient was released from the hospital.

Regular follow up appointments were performed at the Pediatric Clinics at OHSU. On 3-29-82, the patient was seen in the Genetics Clinic at the Child Development and Rehabilitation Center (CDRC), OHSU. A full examination was performed and a differential diagnosis of Treacher Collins Syndrome and CHARGE association was given. However, the diagnosis favored Treacher Collins Syndrome due to the appearance of the ear, mild macrognathia, colobomata of her left lower lid and mild hypotrichosis of the medial portion of her lower lid. Test results revealed no evidence of chromosome involvement. There was no history of other family members with congenital anomalies. A follow up in one year was planned.

Audiology testing was performed on 5-5-82, Results of the test revealed a bilateral hearing loss of severe to profound. This was followed by a thorough ENT examination. It was noted that the structures of the inner ear and the lateral semicircular canals were deformed. The final assessment was a congenital anomaly of the inner ear, possibly the middle ear and possibly the right external auditory canal. Multiple follow up appointments were made with audiology to re-evaluate hearing ability and possibly to fit a hearing aid.

The patients course of development continued uneventful until 11-3-83 when she was admitted with a chief complaint of obstructive apnea. The mother reported that 2-3 weeks prior to admission, she noticed an incident of perioral cyanosis. This lasted three hours. However, during the incident, she was afebrile and in no respiratory distress. Her mother did notice that S.J. awoke often from sleep, snored loudly, and suffered chronic fatigue, but had good energy reserve. In addition, she handled food poorly and was constantly gagging. When she vomited, emesis came from both nose and mouth.

A short sleep study revealed a seven second obstructive event. It was unsure whether she breathes from her nose, but it appears she is an obvious mouth breather. Oropharyngeal surgery was planned to alleviate the apnea.

On 1-19-84 a tonsillectomy, nasopharyngoscopy and partial resection of the soft palate was planned. Again,

intubation was difficult for anesthesiology. ENT first examined the nasopharynx and concluded that the tonsils and soft palate were within the normal range of development and was not the cause of obstruction. No surgical procedure was done. It was recommended that positioning the patient at a 45 degree angle with pillows during sleep, or having her sleep on her side would correct the apnea.

Routine follow up appointments continued at OHSU. It was not until January of 1987 that she was again seen at the Genetics Clinic at CDRC. At this time, it was noted that there was a mild level of developmental delay due to her deafness and blindness in one eye. A hearing aid was fitted and given to her, but she was unable to keep it in. A differential diagnosis of acrofacial dysostosis of Nager and de Reynier was added

Physical examination revealed a weight of 14.2 kg,. height of 102.3 cm, and head circumference of 47.4 cm.. Polotomography of the temporal bones was performed. Results showed abnormal internal ear structures with decreased coils of the chochlea, enlarged vestibule, deformed lateral semicircular canals, shortened and widened anterior canals, possibly misplaced internal auditory canals, small middle ear cavity with displaced ossicles on the right. There is profound deafness. She is enrolled in a special school for the hearing impaired and is doing well with sign language.

Clinical features noted were blepharophimosis of the eye, mild slant of the palpebral fissures and phimosis

of the lateral portion of the eye slit. Eyelashes were dark. There was also a prominent nose and flattening of the malar emminence bilaterally with some areas of scarring. Since the child was doing well medically and socially, a follow up appointment was scheduled in one year.

It was not until 1990 when the patient appeared again with the complaint of "gunky eye". Ophthalmology concluded after examination, that there was a congenital abscence of the puncta in the left eye. This prevented her tears from draining properly. On 6-28-90, a conjunctivodacrocystorhinostomy was performed under general anesthesia. A Jones tube was placed from the left eye connecting into the nose allowing better drainage. Migration of the tube discovered in October of 1990 required a second surgery to replace the tube.

Most recently, the patient was seen for a full updated examination at the Special Craniofacial Disorders Clinic at CDRC in April of 1992. This exam was requested by her school. It was noted here, that the mother is single and seperated from S.J.'s father. She has had a subsequent phenotypically normal daughter by another partner. In the last four years, she has had a new boyfriend, who now lives with them. Chief concerns were that S.J. was not progressing well in school and is now functioning at the first grade level. At school, she would frequently get into fights with others who teased her. Many times, she would initiate the fight.

However, she communicates well using sign language. The issue of low self esteem was a concern. It was suggested that all family members learn sign language. Also, she should be enrolled in recreational activities for the hearing impaired. These activities would hopefully improve her self esteem.

Reports from Genetics now indicate she may have an autosomal reccessive form of Treacher Collins Syndrome. Her features were described as very similar to Treacher Collins Syndrome, but not identical to it.

Physical examination revealed a height of 48 inches, weight of 50 lbs. This falls short of the normal range of development for her age. She is expected to be no taller than 60 inches. No new facial characteristics were reported. It was suggested that she may be a candidate for choclear implants. An MRI was scheduled. A follow up at CDRC was requested in one year.

In June of 1992 , an MRI was performed at OHSU to evaluate her ability to obtain choclear implants. Results revealed many bones, nerves and blood vessel displaced or deformed. Other important findings were that the facial nerve was positioned bilaterally and the carotid arteries course laterally to the cochlea.. With this information, it was concluded that cochlear implants were contraindicated.

Dental Summary

S.J.'s first dental visit was at the age of 2 years on 10-10-83. An examination was performed at CDRC . It

was noted that she had a small mandible. Her tooth eruption was slightly delayed, but her dentition was otherwise normal. The hard palate was high and narrow but intact. Also of importance was a bifid uvula. Dental prophylaxis was performed, oral hygiene instructions were given to the mother, and a one year follow up was planned.

The patient was seen again for dental work in September of 1986. Exam revealed nine primary teeth with caries. All primary teeth were present and the dentition was normal. There was some crowding. The exam was difficult as the child was uncooperative.

The nine decayed teeth were restored on two appointments. In office, oral sedation was necessary with the use of a combination of chloral hydrate, hydroxozine and meperidine for both appointments. During these visits, it was noted that the child snored during the procedure but would awaken often and resist at times. The sedation was considered adequate to complete the needed work. After completion of the treatment plan, the patient was asked to return for a six months recall appointment.

S.J. continued her dental care at the Hospital Dental Service at OHSU in 1989. Again, crowded teeth were noted with poor oral hygiene. Decay was found on seven teeth. One operative appointment was attempted but she was not able to cooperate for the treatment. Dental work under general anesthesia was planned. However, after

the work was approved by the insurance company, the mother did not follow up with comprehensive care.

It was not until April of 1992 that the mother again decided to seek dental care. This time S.J. appeared at the Graduate Pediatric Dental Clinic at the School of Dentistry, OHSU. She appeared to be a very pleasant, happy, playful child. She was able to read lips and understand instructions easily. It was obvious that the patient had a small mouth opening. Vertical opening was measured at 25 mm (normal = 40). Four periapical radiographs of posterior teeth were taken using pediatric size dental film.

Clinical exam was performed revealing a small mandible with crowded teeth. Dental caries of moderate size were found on all four first permanent molars. Severe caries were found on the upper left first and second primary molars. Radiographs revealed that the roots of these teeth were actually resorbing due to the eruption of the permanent bicuspids. Eruption pattern and dentition were both normal. There was moderate plaque accumulation, especially in the molar region, with generalized mild gingivitis.

A prophylaxis was performed, followed by fluoride gel treatment and oral hygiene instructions. It was noticed during the examination and cleaning that the patient could not keep her mouth open for a long period of time. This was due to her obstructed nasal passage and her predominantly being a mouth breather.

A treatment plan of four occlusal and buccal surface alloy restorations on all first permanent molars was planned. This was to be done using local anesthetic only. An orthodontic evaluation was requested once all restorative treatment was completed.

On 4-15-92, she presented to the dental clinic for restoration of teeth #14 and #19. She was seated at a 45 degree angle, and 1.8 cc of 2% lidocaine with 1: 100,000 epinephrine was administered in the upper and lower left quadrants. Due to her extremely small and narrow mouth opening, and her respiratory problems, use of a rubber dam was not possible. The teeth were isolated with cotton rolls and caries excavation was started on tooth #14. A slow speed handpiece was used to remove decay and it was quickly noticed that adequate anesthesia was not obtained. A dose of .9 cc. of the same local anesthetic was administered. After waiting 10 minutes, work was resumed and she still did not have profound anesthesia. Caries removal was continued anyway, and although difficult, the work was completed with the cooperation of the child. Short periods of rest were needed often to allow her to breath.

At this time , it was decided that we would not attempt to restore #19 during this visit, but instead, reappoint her. It appeared that short appointments were desirable.

On 4-27-92, she was seen again to restore the occlusal buccal surfaces of tooth #3. She was seated at 45 degrees and 1.8 cc. of 2% lidocaine with 1:100,000

epinephrine was administered. After waiting 10 minutes, the teeth were isolated and caries removal was started. Again, profound anesthesia was not obtained. A dose of 1.8 cc of the same local anesthetic was used again, and a 10 minute waiting period was given. Anesthesia was still not adequate. Although painful and difficult, the child sat through the remainder of the appointment. A base was placed, and amalgam was used as a final restoration. A small area of caries, on the occlusal (distal) portion of the tooth #3 , was not completed.

On 5-15-92, she returned to complete the restoration of tooth#3, and for a sealant on #30. Again, 1.8 cc. of 2% lidocaine with 1:100,000 epinephrine was administered near #3. After waiting 10 minutes, caries removal was quickly performed. Amalgam was placed as a final restoration. Again, little anesthesia was obtained. The occlusal surface of tooth #30 was restored with a sealant. This procedure was difficult for her because she had to continuously keep her mouth open for a long time to etch and cure the tooth. Treatment was completed, and she was reappointed for her last visit on 5-22-92.

She appeared for her last appointment, to restore the buccal surface of #19. Again, 1.8 cc. of 2% lidocaine with 1:100,000 epinephrine was used. After waiting 10 minutes, decay was removed. As always, no anesthesia was obtained. She cried during the procedure, but was otherwise very cooperative. Amalgam was placed as a

final restoration. After completion of her treatment, she was placed on a 3 month recall.

On 5-24-92, an orthodontic evaluation with a panorex radiograph was performed. Results revealed a Class III right side and a Class I left side molar relationship. Crowded teeth produced a 10 mm. deficiency in the maxillary and the mandibular arches. Tooth #23 erupted completely lingual to the other mandibular incisor. Her incisors were in end-end occlusion. The midlines were even. Eruption pattern was normal. All teeth were present except for third molars. The maxillary canines were underdeveloped and impacted. Hypoplastic mandible and maxilla were again noted. One problem which concerned the mother was that S.J. had a difficult time eating and chewing her food. She takes up to one hour to finish her dinner.

Several recommendations were made. First, it was decided that her teeth and occlusion were functional as they were. Difficulty in chewing was caused by her difficulty in breathing. However, esthetics and function could be improved with orthodontic treatment, followed by maxillary and mandibular advancement surgery. This would be done with a LeForte I procedure and a bilateral split osteotomy.
Further evaluation with an orthodontist and a plastic surgeon who specializes in this area was requested.

S.J. returned for a recall visit on 8-21-92. No new decay was found. Tooth #j, which was previously carious, had now exfoliated. Tooth #13 was now actively

erupting in its place. Other primary molars were loose, and would appear to exfoliate soon. Oral hygiene appeared to be better. However, the mother reported that S.J. brushed her teeth prior to the dental appointment, but does not consistently do it at home. An oral prophylaxis was performed followed by a fluoride treatment and oral hygiene instructions. S.J. was her playful, happy self, and she cooperated as always. Another recall visit was scheduled for three months.

DISCUSSION

The constellation of anomalies seen in Treacher Collins Syndrome leads to complex medical problems. Medical treatment is lifelong and requires attention from several different areas of medical specialty. Diagnosis and treatment begins early, often immediately after birth. Emphasis is first placed in stabilizing the child and managing any problems that may be life threatening. Once this is accomplished, diagnosis and assessment of each individual problem may be performed so that proper treatment can be accomplished through short-term and long-term goal setting.

The first problem one may encounter is poor respiration due to choanal atresia. In the above case, the patient had difficulty breathing from the start of life and nasal stents were needed to maintain a proper airway. With the patient being pre-mature and low birth weight, this lead to additional complications. It is

normal for patients such as this to spend a considerable amount of time in an intensive care unit as she did.

Once this airway problem is managed, other respiratory problems may occur as the child gets older. Sleep apnea is commonly seen in children with craniofacial anomalies. This is due to several anatomical factors. First, difficulty breathing may be caused by pharyngeal hypoplasia. Techniques such as cineradiography, fiberoptic bronchoscopy, nasopharyngoscopy, C.T. scans and lateral cephalometry have all been used to assist in diagnosis. These diagnostic aids will confirm a narrowing of the pharynx throughout the entire vertical height, with reduction through the lateral and anterioposterior dimensions [18,19,20]. Johnstun et al. [19] also noted that sleep apnea may also be caused by a more posterior position of the mandible in patients with micrognathia. As the mandible closes and rests, the normal size tongue moves posteriorly, blocking the pharyngeal airway.

The initial diagnosis of sleep apnea can be made through observation. Some common characteristics are loud snoring, excessive daytime sleep, chronic fatigue, poor night time sleeps, morning headaches and uresis. In schoolchildren, intellectual and personality changes can be seen. Poor attention span may alert the teacher that a problem exists. A teacher may conclude that there is a behavioral problem, or learning when, in actuality, the problems are due to features of the genetic syndrome (eg. deafness).

The two forms of sleep apnea are obstructive and central apnea. Obstructive apnea occurs secondary to pharyngeal or upper airway obstruction. Central apnea occurs when there is a total lack of respiratory effort. They can occur alone, or in combination with one another.

Treatment for patients with sleep apnea have been variable depending on the cause. Maxillary and mandibular advancement surgeries have been used to clear obstruction of the tongue. Pharyngeal flaps and tonsillectomies have also been used to improve the airway.

Pharyngeal hypoplasia may also lead to complications with anesthesiology. Difficult intubations may occur due to microstomia, minimal mandibular excursion, high arched palate, and anteriorly displaced larynx. Airway establishment is important prior to intubation. Patients are often kept intubated after surgery until bleeding and edema from the soft tissue and larynx are under control.

Obstructive sleep apnea combined with choanal atresia can also lead to eating problems. When food is in the mouth, the tongue moves posteriorly thereby blocking off the airway in someone who is micrognathic. A person with choanal atresia breaths through the mouth as the primary source of respiratory flow. Food will then serve as an obstruction to their primary airway. It is common for these individuals to take a long time, up to one and one half hours, to finish a

meal. It would be helpful to encourage small bites of food to make mastication easier so that food spends less time in the mouth. Health professionals may mistake this problem to be a functional orthodontic problem due to crowded teeth. A consultation by an orthodontist would be important in these cases to evaluate such a problem.

Although people with Treacher Collins Syndrome have normal intelligence, hearing, visual and speech problems may delay learning. If this syndrome is not correctly diagnosed, a person may be mislabeled mentally retarded or developmentally delayed. Depending on the severity of these sensory problems, some individuals may have to be educated in special-need programs. Additional delay in the educational process will occur when a child needs to take time off from school for medical treatment. Because they have a normal level of intelligence, these problems can be very frustrating.

Problems with sensory perception as well as attending special schools can lead to a delay in learning social skills. Growth is rapid in the primary school age years and social skills are learned through spending time with peers in the classroom, as well as in the playground. It is normal to see a varying degree of immaturity in these children due to lack of peer association.

The unusual facial appearance may also lead to problems with self-concept. Facial appearance is

important in terms of how one sees themselves as well as how others react to them. Jones[21] reported that children with cleft lip and palate had a lower self-concept than non-cleft people regardless of there sex. Parents of these children believed that teasing increased at school and this also affected school progress. A strong network of support is therefore very important, especially from teachers, counselors and family members.

Surgery to correct facial disfigurement will often improve the self-concept of these children. Timing however is very important because the medical needs of the child must always be a first concern. Lefebvre et al. [22] recommends that surgery should be performed ideally before the school age years, when children are subjected to teasing. However, of greater importance is the readiness of the parent and the realistic expectations of both the parent and the child.

Dental management of a child with Treacher Collins Syndrome may vary when compared to treating a normal child. Dental caries and plaque control should not be any different. However, if these children are immature, oral hygiene may not be taken seriously. From the parents' perspective, the other medical and social problems may be so overwhelming that oral hygiene may be of secondary importance. Microstomia and crowded teeth may also contribute to poor oral hygiene. It is important that oral hygiene be emphasized with both the parent and the child. Short

recall intervals may help the dentist or hygienist better monitor the oral home care. Small children's size toothbrushes may be needed even as the child gets older, due to microstomia.

Hearing, visual and speech problems may make it very difficult to communicate when treating these children. Pediatric dentists sometimes prefer that parents are not present in the treatment room when procedures are being performed. In cases with impaired sensory loss, it is wise to have the parent or a family member present in the operatory to assist in communication. The dentist must always be aware of the overall needs of the patient and not concentrate only on their teeth. Communicating through facial expression and hand signals will make the procedure less stressful for the child.

Maintaining a proper airway is of vital importance during operative procedures. If the child is a mouth breather due to choanal atresia and also obstructs easily, the procedures in the mouth will compromise their airway. It is helpful that the patient be seated at a 45 degree angle so that the mandible is not forced posteriorly. A rubber dam may be contraindicated because firstly, it may be difficult to get one on in a small mouth and secondly, because it will block off the primary airway. Working in the mouth also interrupts breathing. It is wise to take breaks often to let the child catch a breath. A single appointment may require much physical exertion from the patient. Multiple, short

appointments instead of single, long procedures will increase patient compliance.

Local anesthesia may be difficult to achieve due to the misplacement of the nerves. Herring et al[23] studied skulls of individuals with mandibulofacial dysostosis. She found similar bony structures among each subject, but a wide variation in pathways of nerves and vessels. One may not expect to achieve profound anesthesia at each visit. For patients who are difficult to anesthetize, working quickly to complete a procedure may be your only choice. If the treatment plan is extensive, dental procedures under general anesthesia may be a final option.

Once permanent teeth erupt, orthodontic treatment may be required to alleviate crowding. Consultations should be performed in conjunction with a plastic surgeon or oral surgeon because surgical correction is often needed. This surgery will also help in eliminating sleep apnea. Past studies have shown a decrease in respiratory disturbances following maxillo-mandibular surgery in the patient with obstructive sleep apnea. If this surgery is done in conjunction with orthodontic treatment, it may be performed during the teenage years when most of the permanent teeth have erupted and facial growth has leveled off. Treatment planning should be done in the mixed dentition phase, well ahead of the time of surgery.

Finally, making a definitive diagnosis of any genetic anomaly can often be very difficult. Several

factors must be considered in the process. Ideally, a pattern of inheritence should be established through careful documentation of a multi-generational family history. Unfortunatley, factors, such as new mutation, imcomplete penetrance and variable expressivity, make this difficult.

Clinical characteristics are very important in the diagnosis of a genetic disorder. There are usually one or two dominant features, along with several other characteristics that may exist. Variability in features sometimes will complicate a diganosis. A certain number of clincial features must be present before the final diagnosis of a syndrome can be made.

Once a specific syndrome is suspected, chromosomal, biochemical or molecular testing may be available to confirm the diagnosis. However, as in the case of Treacher Collins syndrome, many genetic disorders have not yet been characterized at the gene or chromosome levels. Combining family history, genetic mapping, and clinical features may be difficult but can also be exciting when diagnosing anomalies. In S.J.'s case, it is likely but not competely certain that she has Treacher Collins syndrome. Even without a definitive diagnosis, having an idea of what it might be is helpful when dealing with the complex problems associated with anomalies.

Figure 1: Front view. Most noticable in this view is the ears bending anteriorly.

Figure 2: Right side profile depicting the characteristic mandibular hypoplasia. Also noticable is swirling of the hair or sometimes described as "tongue shaped" growth of hair which extends to the cheek.

Figure 3: Left side profile again showing the mandibular hypoplasia. The scar anterior to the ear has been present since birth.

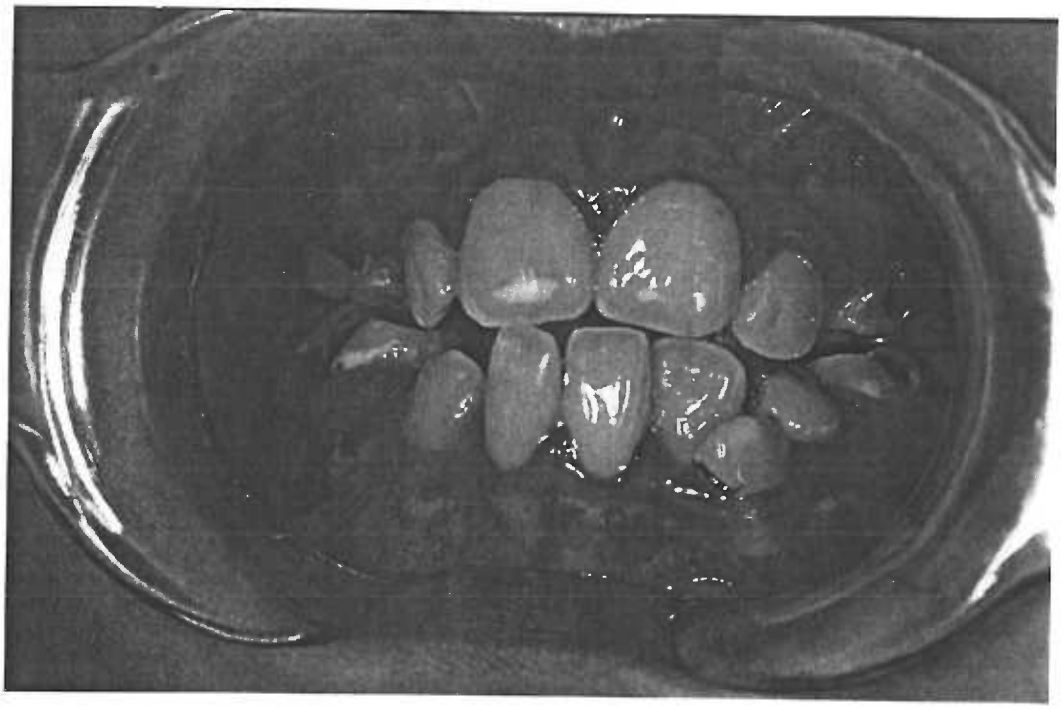

Figure 4: Dentition in centric occlusion showing the severe crowding and an anterior end-end occlusal relationship.

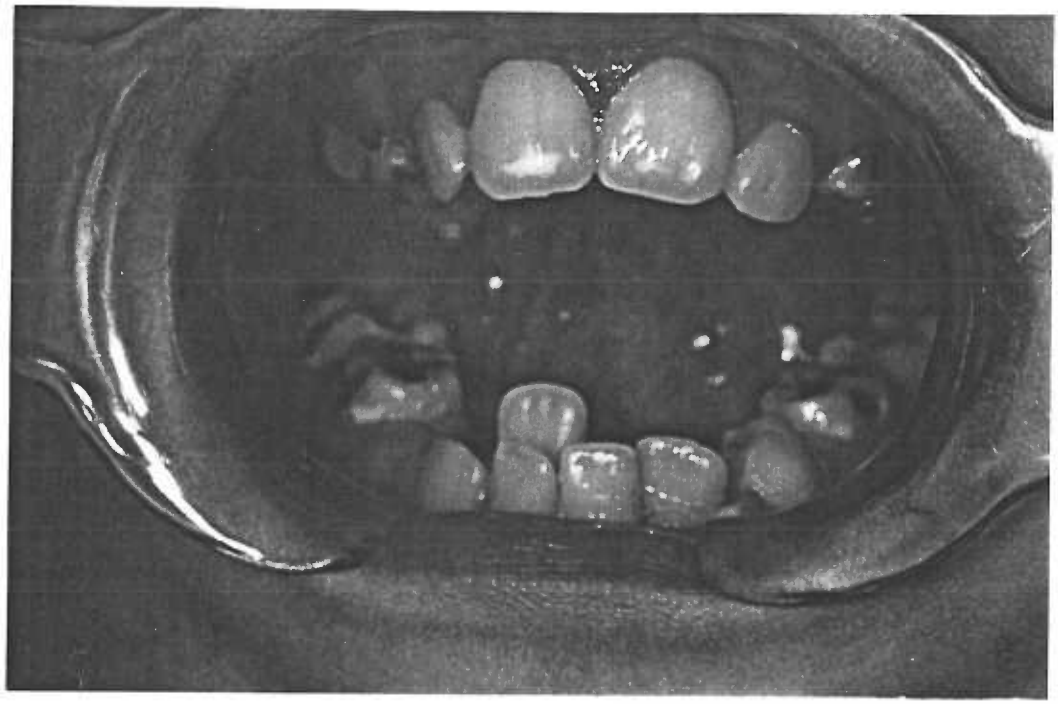

Figure 5: Vertical opening was measured 25 mm.. This view also shows tooth #23 which erupted lingual to the other mandibular incisors due to lack of space.

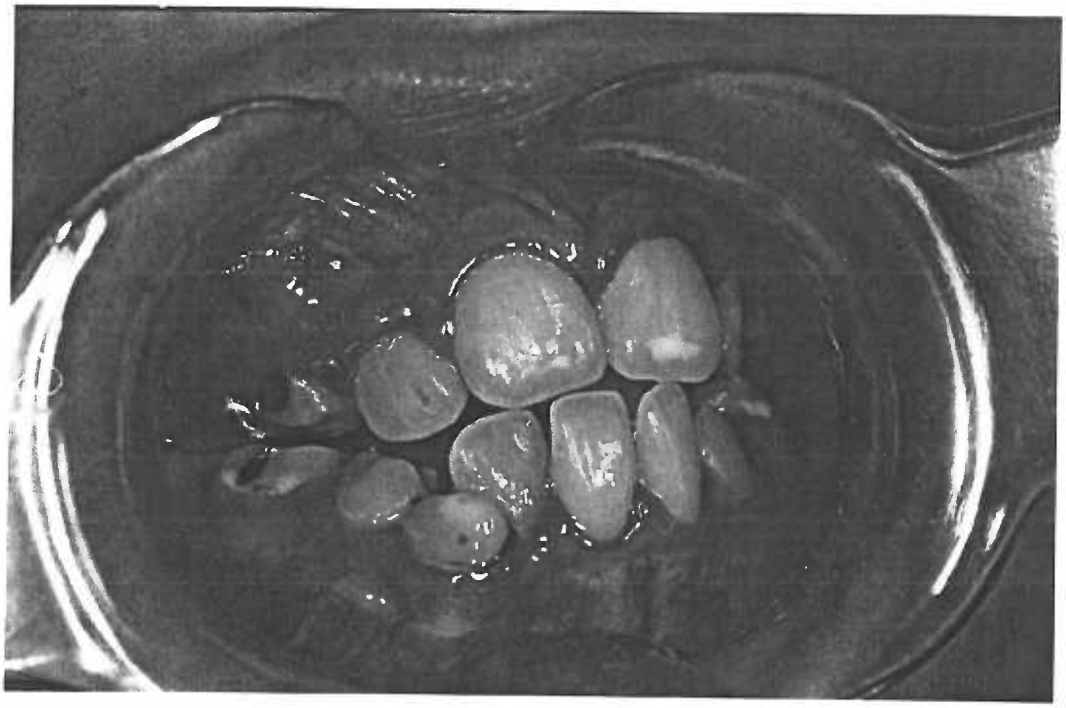

Figure 6: The right view of occlusion was described as Class III. Tooth #27 is erupting labially to #26 due to limited available space.

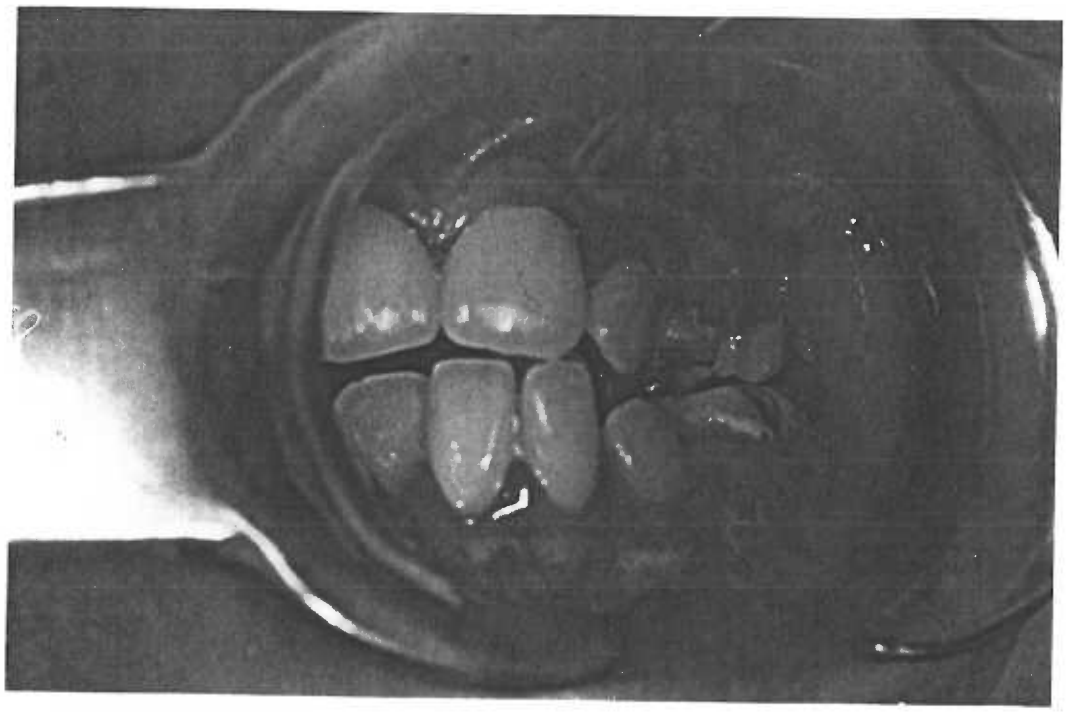

Figure 7: The left side occlusion was described as Class I. Notice the areas of decay in the upper and lower molar regions.

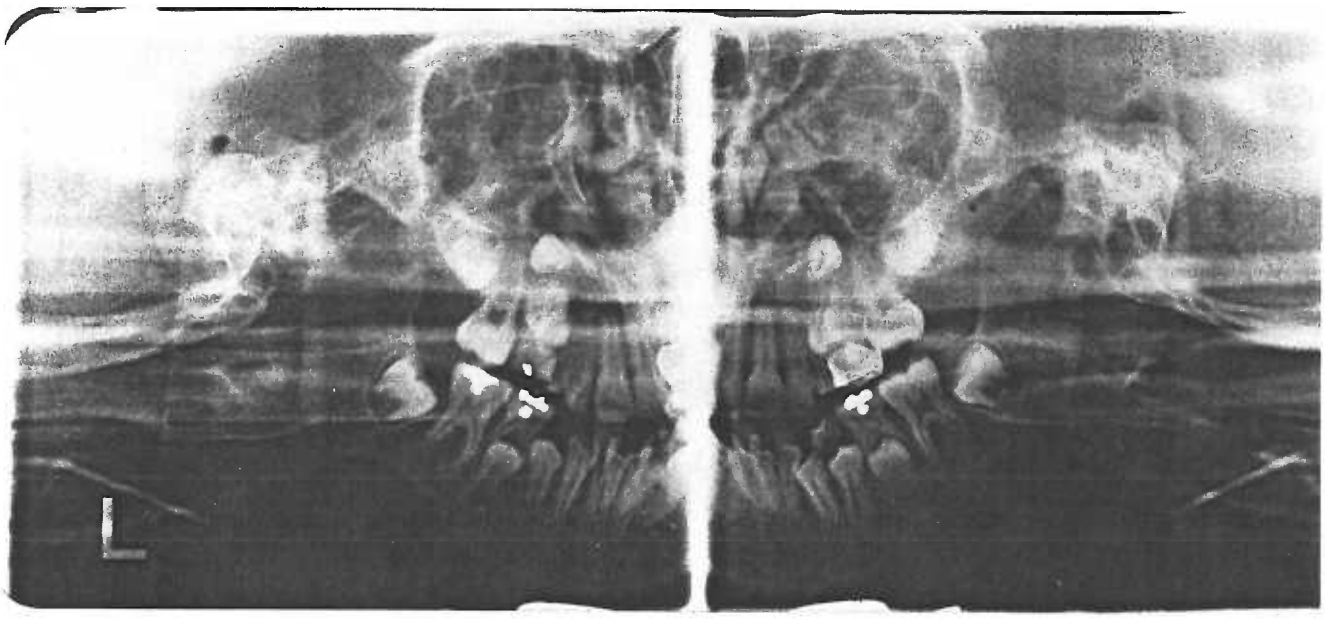

Figure 8: Panoramic radiograph showing the severe crowding. The small follicles appearing in the upper premolar regions are the cuspids.

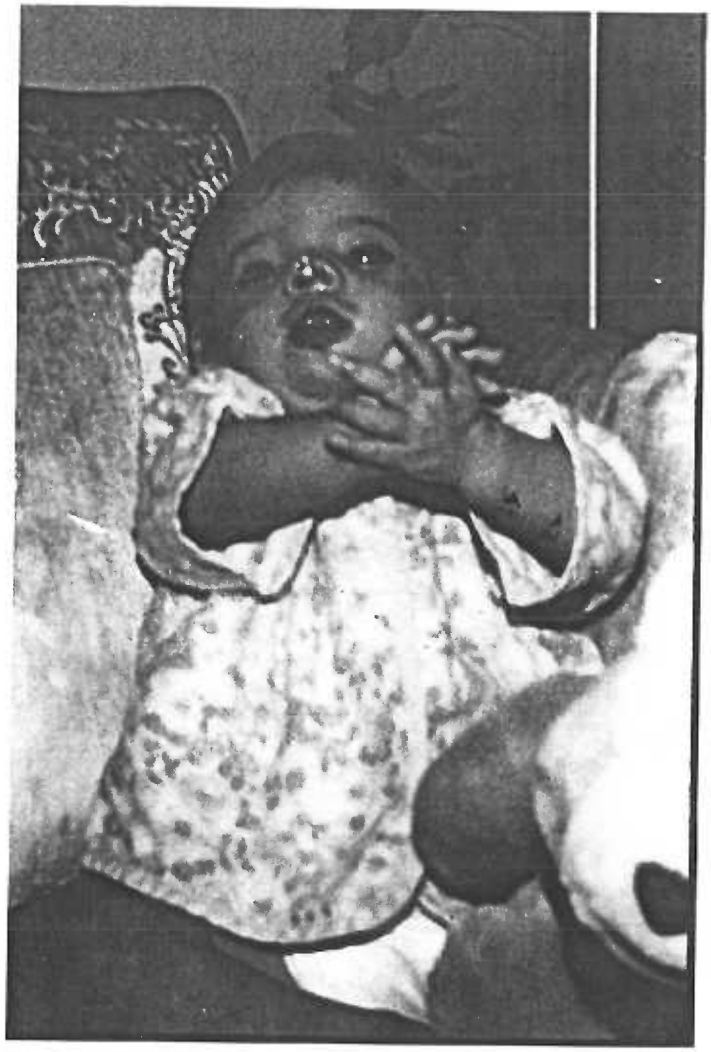

Figure 9: S.J. at age 3 months wearing the nasal stents which helped her to breath.

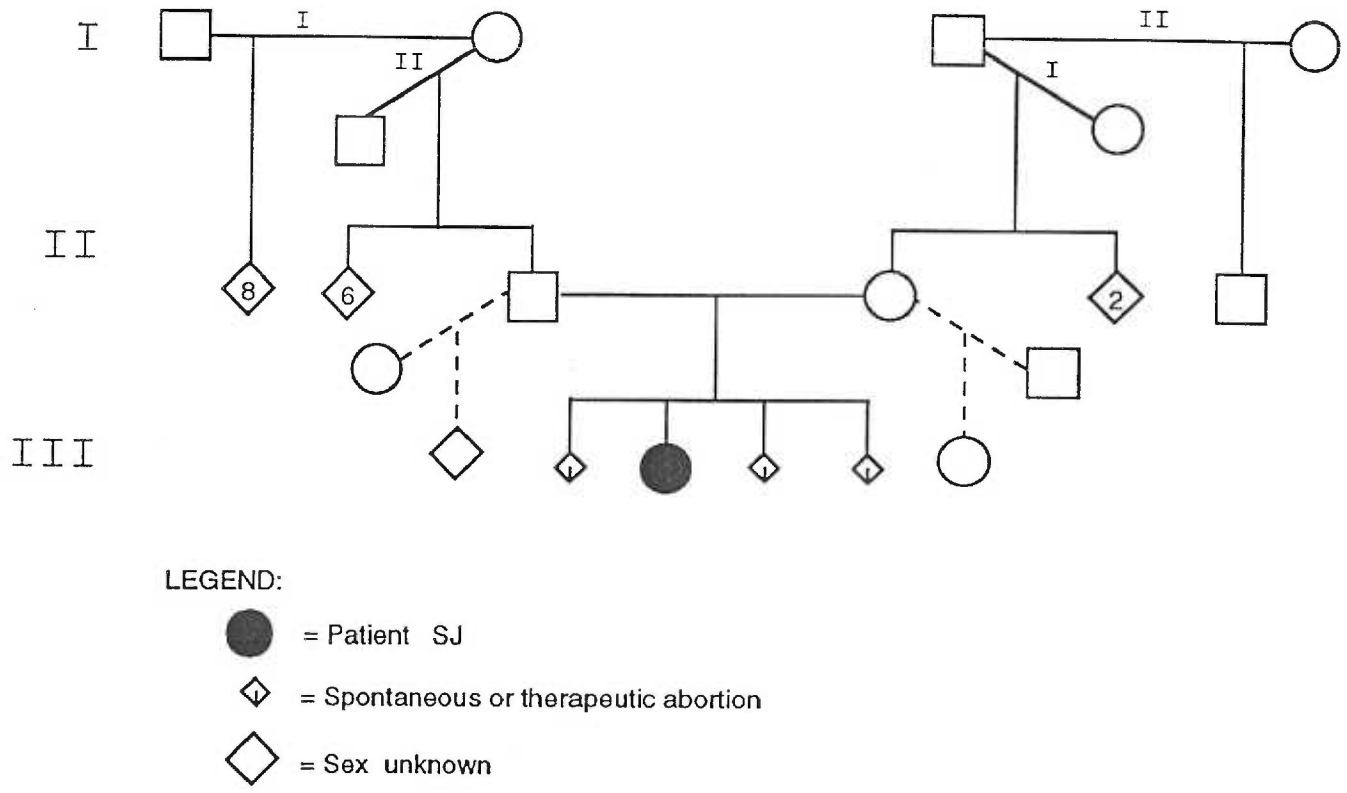

Figure 10: Pedigree showing no additional affected members in three generations.

Antimongoloid slanting palpebral fissures	89%
Malar hypolpalasia, +/- cleft in zygomatic bone	81%
Mandibular hypoplasia	78%
Lower lid coloboma	69%
Partial to total abscence of lower eyelashes	53%
Malformation of auricles	77%
External ear canal defect	36%
Conductive deafness	40%
Cleft palate	28%
Incompetant soft palate	32%
Projection of scalp hair onto lateral cheek	26%

Table 1 Incidence of abnormalities associated with Treacher Collins syndrome. (Taken from Smiths Recognizable Patterns of Human Malformations, K.L. Jones, 1988)

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