STICKLER SYNDROME
SUPPORT GROUP
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ORO-FACIAL ABNORMALITIES WITHIN STICKLER SYNDROME

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1. ORO-FACIAL ABNORMALITIES WITHIN STICKLER SYNDROME

- Both Type 1 and Type 2 Stickler Syndrome patients have oro-facial abnormalities to some degree which will be one or more of the following:
- A full cleft palate, a submucous and/or bifid uvula, or a high-arched palate. Cleft lip is not a symptom of Stickler Syndrome, and therefore is extremely rare.
- Micrognathia, where one or both jaws are unusually small, resulting in poor contact between the chewing surfaces of the upper and lower teeth.
- Glossoptosis – a tendency for the tongue to 'ball-up and fall backwards towards the throat, which could block the airway.
- These symptoms are similar to those found in Pierre Robin Sequence (PRS) and it has been reported that around 30% of children diagnosed with PRS are later diagnosed as having Stickler Syndrome.
- Facial characteristics include a flat face with a small button nose and no nasal bridge, epicanthic fold, and large prominent eyes. However, the appearance does tend to improve, especially by the time the child starts school.

2. CLEFT INVOLVEMENT WITHIN STICKLER SYNDROME

It is interesting to note that a cleft palate is the fourth most common birth defect, occurring in one in every 700 live births. It is no one's fault that a child has a cleft palate, but a quirk of nature that is beyond our control. It is known that the majority of clefts are either of genetic origin or occur for environmental reasons, or a combination of both. A cleft is formed by the 4^{th} - 8^{th} week of pregnancy, before many women are even aware that they are pregnant, and

occurs when the tissues of the face fail to come together, leaving a gap - a cleft. Cleft palates range from a simple notch of the soft palate at the very back of the mouth to a far more extensive involvement of both soft and hard (the roof of the mouth) palate. It is a repairable birth defect, although feeding the baby can be difficult at first (see section on feeding).
For families who have a history of Stickler Syndrome, as well as families who have no history of Stickler Syndrome but are suspected of having the condition, a cleft palate may be the only visible sign that anything is wrong.

2.1. WHAT HAPPENS NEXT?

- Once a cleft abnormality is discovered you will meet a team of medical professionals - called a Cleft Palate Team. This basic team will consist of a Paediatrician, a Surgeon who repairs clefts, an Audiologist, a Speech and Language Therapist, and an Orthodontist. They will consult with each other and plan how best to help the baby. This association with the team will last into adulthood. As cleft palates are sometimes associated with syndromes, such as Stickler Syndrome, the child's eyes should also be checked as a matter of routine.
- Children with Stickler Syndrome usually come to the attention of the medical profession because there is a family history of Stickler Syndrome and are therefore diagnosed at birth. However, where there is no family history as in most cases with a cleft involvement, they are initially diagnosed at birth as having Pierre Robin sequence. This is because Pierre Robin sequence has the same three possible features as Stickler Syndrome, which may or may not be present - micrognathia, glossoptosis, and a cleft palate.
- The face of a new-born 'Stickler' baby may have characteristic features. These can include midfacial hypoplasia - where the middle part of the face (including the upper jaw) has not developed far enough forwards

and downwards. Other features may include prominent eyes, a vertical fold of skin from the upper eyelid that covers the inner corner of the eye, and a small button nose with little or no nasal bridge. This distinct characteristic 'Stickler' face can lead consultants to suspect Stickler Syndrome and go on to correctly diagnose the condition Many of these features, especially the short lower jaw, improve with time so that the facial abnormalities can look less prominent by the time the child starts school. However, if the abnormalities are severe, facial surgery with nasal reconstruction may be required.

2.2. CLEFT PALATE

- Although timing of a cleft palate repair will vary from surgeon to surgeon, most are repaired around the age of 9-12 months. Unfortunately there is no guarantee that this will be the only surgery required and many children with a cleft require revision work to be undertaken as they develop. Your cleft surgeon will give you a better idea of what to expect, as each case is unique.
- Until cleft palate surgery is carried out, your baby may experience feeding problems (see section below on feeding), and again the cleft team will advise on the range of special feeding bottles available and offer appropriate support. Parent support groups such as CLAPA are also only too happy to offer support.

2.3. SUBMUCOUS CLEFT

A sub-mucous cleft is one in which the surface or lining of the soft palate (mucosa) is intact (complete), but the muscles beneath the surface have not properly joined. Usually the only outward sign of a sub-mucous cleft is a bifid uvula (see below). Early recognition is important as the muscles cannot work properly and so speech and hearing will be abnormal. Unfortunately it is usually not

detected until a child develops speech difficulties at around the age of three, but is often detected much later following unsuccessful speech therapy. Children born with a sub-mucous cleft do not always need corrective surgery. If the cleft is not severe enough to create problems, a course of intensive speech therapy is usually enough. However, in many cases surgery is necessary to restore muscle continuity in order for the child to achieve the kind of function (control) and hearing needed to aid good speech. Your Cleft Team will advise on the best procedures.

2.4. BIFID UVULA

The uvula is the small soft extension of the soft palate that hangs from the roof of the mouth above the root of the tongue. It is made up of muscle, connective tissue, and mucous membrane. Bifid means split into two, therefore a bifid uvula is a split uvula. It may have an adverse effect on speech but if not, it can be left.

2.5. HIGH ARCHED PALATE

A high arched palate occurs when the palatal shelves, which start at each side of the palate, do not come down as far as they should creating an abnormal shaped palate. There is no cleft palate with a high arch palate. However, this abnormal shape can change the position of the tongue and can cause speech problems.

2.6. FEEDING DIFFICULTIES

- In order to feed, a baby must be able to form a vacuum inside his/her mouth and position the tongue properly. Babies with clefts may not be able to create this vacuum or position their tongue properly. Some have a smaller lower jaw making swallowing difficult. Your Cleft Team will decide on the best feeding plan to suit you and your baby.

- A thin feeding tube may be passed into the baby's stomach though the nose to help those who also have a small jaw. Breast-feeding is rarely possible for those with a cleft palate and a mother should not feel guilty at not being able to feed her baby. Mother's milk can always be expressed. Many babies will need help with feeding by using one of a range of different types of bottles and teats. Your Cleft Team will supply the one best suited to your baby initially, and you will be advised where to buy on-going equipment. In some cases, and in order to help with feeding, an Orthodontist may be asked to provide a small dental plate, although there is little evidence to support the theory that they really help with feeding. In some cases a plate may be used to keep the tongue out of the cleft palate prior to repair. Babies with clefts may swallow more air than normal during feeding, especially if the flow of milk is either too slow or too fast. If this happens, it may show by the baby having a blue moustache, being extra sleepy or bringing up some of the feed. If this happens, stop two or three times during a feed to burp the baby, or try sitting the baby in a more upright position. **Every mother and baby is unique** so it is impossible to give hard and fast rules about feeding. The best method is the one that suits mother and baby. Try the simple things first, and relax. Allow the baby time with one method before deciding it is not working and before trying alternatives. The secret of success is to take your time, be patient and calm. Also remember that babies can lose up to 10% of their birth weight, but will usually regain it in two to three weeks. Babies with a cleft palate may take longer to gain weight.

3. HEARING LOSS WITHIN STICKLER SYNDROME

There are basically two different types of hearing loss, known as conductive and sensorineural. Individuals with Stickler Syndrome can be affected by either or both. In a child a persistent hearing loss can affect the development of speech and language. In an adult a persistent hearing loss may affect the individuals ability to communicate, and thus limit social and professional opportunities. There are many treatments and strategies for dealing with hearing loss that are readily available.

3.1. CONDUCTIVE HEARING LOSS

A conductive hearing loss involves the sound conduction mechanism, that is the eardrum and the bones of hearing (ossicles) within the middle ear. The hearing loss is of a moderate extent. Conductive hearing loss is very common in Stickler Syndrome. The most common cause of a conductive hearing loss, especially in childhood, is Otitis Media (sticky fluid causing pressure behind the eardrums). This can be acute or chronic, and is very common in children with Stickler Syndrome. It is often called glue ear and is due to a malfunction of the Eustachian Tube. The symptoms of Otitis Media, or other ear infections, can vary from a high fever and lethargy to no physical signs at all. If a child's behaviour changes dramatically - irritable, fussy, etc – think ears! Remember that a well child can develop a full-blown ear infection in just a few hours. A more unusual cause of a conductive hearing loss is a malformation of the ossicles. Whilst this has been reported in Stickler Syndrome, it does not seem to be common.

3.2. TREATMENT FOR OTITIS MEDIA

If the problem persists the consultant may decide to perform a minor procedure under general anaesthesia. A tiny incision (called a myringotomy) will be made in the

eardrum, the purpose of which is to remove the fluid behind the eardrums and to ventilate the middle ear. Sometimes to make these incisions more effective, tiny tubes (grommets) are inserted - a myringotomy with tubes. These tubes are made of a rigid plastic or metal material, and remain in the ear for between a couple of months to a year or more. However, due to the tympanic membrane or eardrum of people with Stickler Syndrome being thinner and more mobile than in the general population, they respond less well to having grommets fitted. Eventually the tube will fall out by itself and, hopefully by this time, the child's Eustachian tube will function normally. If there is still a problem with infections and pressure, another set of tubes may be inserted to replace the first. If the condition still persists after several sets of grommets, the surgeon may decide to insert a more permanent set of tubes to reduce the eventual scarring of the eardrum. These more permanent tubes are called 'T' tubes and are made of silastic - a very soft synthetic rubber. They are shaped like a letter T, with the wings of the T folded flat. They are inserted through the eardrum during surgery and pop into place, opening the wings of the 'T' against the inside of the eardrum. As these are held tightly in position behind the eardrum they are usually surgically removed. After care is important and the consultant will advise. However one important point to remember is that the child's eardrums must be protected from water. Although there is some controversy concerning the over-use of tubes in young children, it is important to remember that all cleft-affected children have an anatomic deformity that reduces the ability of the ear to function normally. This has an effect not only on hearing but also speech development. In some cases of conductive hearing loss the consultant may advise that a hearing aid is a safe and effective alternative to surgery.

3.3. SENSORINEURAL HEARING LOSS

Sensorineural hearing loss or 'nerve deafness' is associated with reduced sensitivity of the inner ear to sound. Sensorineural hearing loss in Stickler Syndrome is less common than a conductive loss, but may still affect up to 40% of individuals. It is possible to have both a conductive and a sensorineural component to a hearing loss: this is called a ‘mixed ‘hearing loss.

3.4. TREATMENT FOR SENSORINEURAL HEARING LOSS

Sensorineural hearing loss can be managed with hearing aids and therapy if it is causing a problem with education, social or professional activities. It is encouraging to note that the quality of hearing aids has improved considerably over the last few years particularly with the introduction of digital aids. These more sophisticated modern hearing aids are now available on the NHS.

3.5. HEARING LOSS IN ADULTS AND CHILDREN WITH STICKLER SYNDROME

Any baby born with a cleft palate or a family history of Stickler Syndrome should be routinely tested for a hearing loss - see hearing tests below. If you have any concerns about your child's hearing, speech, language or development at any time, you should request a referral to your local Audiology service through your GP.

Adults should ask themselves the following list of questions to determine if they need to be assessed for a possible hearing loss:

- Do you feel people are mumbling or not speaking clearly?
- Are you able to hear people, but not understand what they are saying?
- Do you have trouble understanding women, children's and soft voices?
- Do you have trouble hearing over the telephone?

- Do you need to turn the television up louder than other people in the household?
- Do you find yourself having to ask people to repeat themselves?
- Do you have trouble following a conversation in a noisy room?
- Do you have ringing in your ears (tinnitus)?

If the answer is yes to any of the above questions then you should be thinking about seeking an assessment.

3.6. HEARING TESTS IN CHILDREN

- Any child diagnosed as having Stickler Syndrome or Pierre Robin sequence should be referred to an Audiology Service for a hearing test which takes between 30 minutes and an hour. Different tests are used depending on the age of the child.
- Children up to the age of 8 months of age may be given an otoacoustic emission test. In a normal functioning ear sound waves, called otoacoustic emissions, are sent out from the ear in response to external sound. By placing a tiny specialised microphone in the child's ear canal an audiologist can measure the signals and investigate the child's hearing. To be able to record these very small sounds requires the child to be fairly quiet. This is not always possible in a very lively young baby; so another test called an Auditory Brainstem Response test may be used. When the ear receives a sound, a tiny electrical impulse is sent along the auditory nerve to the brain. The auditory brainstem response test records these nerve impulses, and allows the audiologist to establish at what level the child could hear the sound. To carry out this test a series of click sounds are played into the ear through a headphone. The response of the nerve is measured using recording leads placed on the baby's head. As both these tests record very quiet signals, it is helpful if the child is

relaxed, or better still, asleep. It is a good idea to bring feeding equipment or a comfort blanket or toy along to the appointment if you feel these will help the baby to relax.

- A child between the ages of 8 and 30 months is able to locate a sound source by moving its head and is often tested by means of a visual response audiometry. The child is placed on the parent's lap, and the tester sits in front of the child holding its attention by playing with some toys. A sound is then played from a loudspeaker to the side of the child, and if the child turns to look, a musical toy is used to encourage the child to turn to further sounds. By using this method the audiologist can establish the quietest sound the child can hear. Other tests may also be used. For example a test that looks at the functioning of the eardrum, and the area behind it - the middle ear - may be used. In this test, called a Tympanometry, a small probe is placed in the entrance to the child's ear canal, and plays a sound into the ear while altering the pressure in the ear canal. A microphone then records any sound that is reflected back from the eardrum. This test can help to establish the presence of fluid (glue ear) in the middle ear.
- Adults are usually tested in a soundproof room with an audiometer. The patient is asked to respond when they hear a tone or sounds that come through the headphones. The result is an audiogram. This is a graph showing the results of the hearing test. Low pitch or frequencies (tones) are shown on the left and high frequencies on the right. Soft tones are at the top of the graph and loud sounds at the bottom. The Audiologist will explain the results of these tests, and the types of hearing difficulty will be discussed.

3.7. FACTS CONCERNING HEARING LOSS

The early diagnosis of hearing loss in children is very helpful in the development of speech and language.

Even a mild hearing loss affecting just one ear can hold a child back in school, and the appropriate management of such a loss is advised. A hearing loss can affect the quality of life of an adult, but effective management and therapy are readily available.

4. OTHER DIFFICULTIES ASSOCIATED WITH CLEFT PALATE

4.1. SPEECH

The effect of a cleft palate on the speech of a child is variable. During speech the palate is important for sealing the nose so that sufficient air flows through the mouth to produce good clear consonants and vowel sounds. If the palate is not able to close off the nose adequately, nasal sounding speech (called hypernasal speech) may result. Once the palate has been repaired the majority of children go on to develop clear speech. Some children who may continue to have difficulty with speech production may require further surgery - possibly a pharyngoplasty - if the palate is not closing the nose adequately. See section on *Speech Therapy Within Stickler Syndrome.*

4.2. DENTAL AND ORTHODONTIC CARE

- Children who have cleft problems associated with Stickler Syndrome may have malocclusion - poor contact between the chewing surfaces of the upper and lower teeth. Orthodontic appliances or 'braces' are used to apply gentle pressure to the teeth and jaws with the aim of improving the bite or the alignment of the teeth. This treatment is usually carried out in two stages - around the age of eleven or twelve, soon after the permanent incisors (the cutting teeth) and other second teeth have come through, and sometimes again around the age of sixteen or seventeen, when the majority of the permanent teeth have come through. Early orthodontic treatment is aimed at expanding the dental

arches if they have collapsed inwards, which sometimes happens after palatal surgery and the formation of scar tissue. Misplaced teeth can also be treated at this stage and if there is overcrowding, teeth can be removed to allow the other teeth to erupt in to a better position. Treatment is either with removable appliances or fixed (railway track) appliances depending on the complexity of treatment required. Later orthodontic treatment may be aimed at further improving the dental arches, their relationship to each and the individual position of teeth. Fixed appliances (braces) are invariably used to improve both the alignment and the bite of the teeth with the aim of creating a good dentition - the arrangement of teeth in the mouth. This is important for health, it enables the teeth to function well, and importantly for the adolescent, it is pleasing to the eye. The treatment is generally spread over two years, with monthly or six weekly visits for routine adjustments. This is followed by a retention phase to minimise the possibility of a relapse.

- However, when the middle part of the face has failed to grow forwards, as a result of the cleft, surgery (an osteotomy) to advance the bone and the teeth within it, is required. Orthodontic treatment may be a part of this programme, which usually commences as growth is nearly finished, and is designed to ensure that the teeth meet correctly after the surgery has been completed.

5. SPEECH THERAPY WITHIN STICKLER SYNDROME

In order to speak correctly there must be a good seal between the mouth and the nasal passage. The movement of the soft palate at the back of the mouth, and the movement of the walls of the throat (pharynx) make this seal tight. In individuals with Stickler Syndrome, this seal may be poor if they have a cleft palate or a

submucous cleft palate. If this seal is poor then a child will have difficulty in making the proper sounds of consonants such as p, b, t, d, s, and ch. There may also be a nasal sound to the voice due to air escaping down the nose. A Speech and Language Therapist, who will help the child to speak as well as possible, should routinely check children who experience these problems. You will first meet the Speech Therapist when you attend a Cleft Palate Clinic. When the child is about 9 months old, a formal arrangement will be made for the child's speech and language development to be assessed by the Speech Therapist. Parents should be aware that until a child's palate is completely closed there are certain sounds that the child will be unable to achieve. If there is a nasal sound in the child's voice, the therapist may use special computer equipment to monitor the air passages as the child speaks. Sometimes a tape recorder and/or a video is used. One of the ways of examining the soft palate and the side walls of the throat is to use a moving x-ray called a video-fluoroscopy. Another method used is called a nasopharyngoscopy. This involves lightly anaesthetising, with a spray, one side of the nose and then a thin fibre-optic telescope is passed into position to record the palate and side walls of the pharynx (throat) as they move during speech. Nasendoscopy cannot usually be done in children under the age of six or seven as it requires considerable cooperation. After examination the therapist will design a special programme of activities and sound exercises appropriate to the age and development of the child. The child will be seen again at least annually until five or six years of age. Speech and language will be carefully monitored through to the age of fifteen. These regular assessments will be planned to coincide with primary and secondary surgery, speech development and any later involvement with orthodontic or surgical intervention. If there is a problem that will not improve with speech therapy alone, it may be necessary to

improve the seal at the back of the throat with an operation. The Surgeon will liaise closely with the speech therapist to ensure that he/she has all the information needed. Once the Surgeon understands the particular problem, he or she may then choose to carry out an operation.

6. RE-PALATOPLASTY AND PHARYNGOPLASTY

If the nose is not shut off when we speak, air leaks into the nasal cavity and speech becomes nasal in tone as the air that should be used to make sounds in the mouth escapes through the nose. Sometime this may be due to the soft palate not moving at all, or nor not enough, so it can't make closure against the back of the throat. In this situation, taking the palate apart and starting again, re-palatoplasty, or re-do of palate repair, is the best option. If successful, the palate can then function normally and close off the mouth from the nose. However, there are some instances where, despite normal palatal movement, speech is "nasal". In these cases, pharyngoplasty is performed. This operation changes the shape of the throat to prevent too much air escaping down the nose, thus helping speech. This operation can also pave the way for therapy to improve speech sounds. A small proportion of children who have their cleft palate repaired in infancy may need a pharyngoplasty to help speech. Other children with no obvious cleft, like a submucous cleft, or children who have a very small palate, may also benefit from this operation. There are many different types of pharyngoplasties, and the surgeon will choose the best one for the child's problem. The surgeon may use moving x-ray pictures of the palate and throat or an endoscope (a small telescope) passed through one nostril to view the palate before he/she decides which type of pharnyngoplasty to use. It must be remembered that basically, whichever procedure is used, the operation changes the shape of the throat to help speech.

7. PROBLEMS TO LOOK OUT FOR DURING THE TEENAGE YEARS

- Adolescence is a difficult time for children, but children who have a cleft abnormality may feel self-conscious about their appearance and speech. They may experience feelings of inferiority because they feel different to their peers, and may look and sound different. Parents may need to spend some time with the teenager talking through any worries and concerns and may need to seek professional advice through counselling. Low self-esteem can be a particular problem. Unfortunately, society often has lower expectations both academically and socially of someone who looks and sounds different. The teenager may also have a lower expectation of him or herself if allowed to believe that their peers and teachers have low expectations of them too. Teasing and bullying can be a huge problem for someone who looks different and because of sight and hearing problems the 'Stickler' teenager may find it difficult to make friends and this can lead to withdrawal. On the other hand, in order to assert him or herself the teenager may also become aggressive and disruptive. The child may be regularly absent from school due to medical appointments and surgery and this naturally disrupts his or her social life and creates pressures at school as they can quickly find themselves in a catch-up situation with lessons and homework. Communication can also be a problem. A teacher or peer may misunderstand a child with a cleft problem and this can lead to frustration and bouts of bad behaviour.

8. HOW YOU CAN REACH US

Write to:

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