Center for Cleft lip and Palate
and Craniofacial Malformations
University Hospital Tübingen

Information for Parents
Robin sequence

Universitätsklinikum
Tübingen

Introduction

Robin sequence (RS, also called Pierre-Robin sequence) is a congenital malformation that affects approximately 80 infants per year in Germany. It is characterized by mandibular retrognathia, an obstruction of the upper airway and the tongue falling back into the pharynx (glossoptosis). In 80-90% of cases, the disease is accompanied by a cleft palate.

Due to the complexity of the clinical picture, an interdisciplinary team of experts is essential, which in our center includes neonatology, orthodontics, oral and maxillofacial surgery, pediatric neurosurgery and speech therapy. Other specialist areas such as prenatal medicine, neuropediatrics, anaesthesiology, ophthalmology and ENT medicine, phoniatrics and paediatric audiology, radiology, general dentistry and medical genetics are regularly consulted to provide the best possible care for patients with this rare disease.

The clinical picture

Clinical symptoms.
How do I recognize that our child has a Robin sequence?

As mentioned above, affected patients have a small, displaced lower jaw, a tongue that is displaced into the throat and the resulting difficulties breathing. In addition, there is often a cleft in the palate.

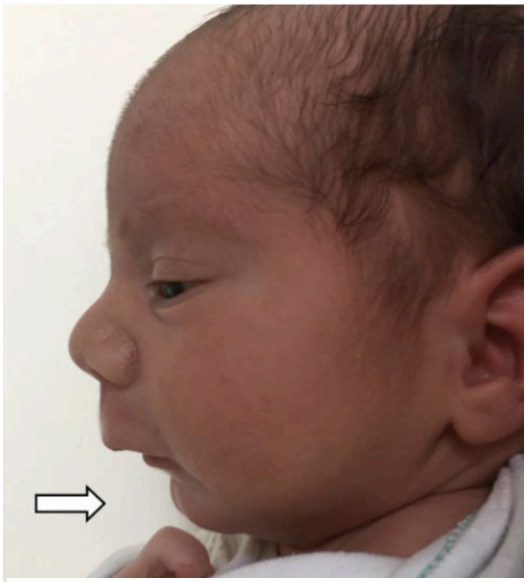

Illustration 1: Child with PRS and pronounced mandibular advancement (arrow)

Due to the anatomical conditions, patients may experience pronounced sucking and drinking problems and a resulting failure to thrive. There is also a risk of a partially or completely obstructed airway and thus strained breathing, respiratory noises or even pauses in breathing (so-called obstructive apneas). These obstructive apneas, which mainly occur during sleep, can lead to disturbed sleep with recurring awakening reactions ("arousals") and oxygen deficiency. The increased work of breathing can further exacerbate feeding and failure to thrive. Mental development can also lag behind and, rarely, even sudden deaths have been reported.

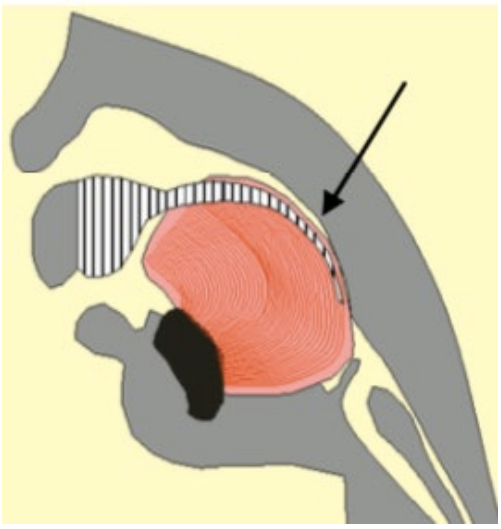

Illustration 2: Airway obstruction in RS due to mandibular retraction and displaced tongue

The varying degrees of severity of the clinical picture lead to a wide variation in clinical symptoms. For example, breathing disorders with the risk of obstructive apneas and oxygen deficiency can be observed in some children at birth, but may also only occur after some days or weeks.

When should families contact us?

Affected families can contact our center at any time with questions and concerns, regardless of the age of their child.

However, the treatment of facial malformations is particularly promising if it begins in the first few days after birth. If a Pierre Robin sequence is already suspected from a prenatal ultrasound or is detected early after birth, we recommend that affected families visit our center as soon as possible after birth.

Treatment Goals

The aim of our treatment concept is

→ to eliminate any respiratory disorder

→ to enable normal food intake with age-appropriate growth

→ if necessary, to close the cleft palate in order to achieve a normal speech development

→ if necessary, to carry out orthodontic treatment in childhood and adolescence in order to achieve a normal subsequent development.

The Tübingen Treatment Concept

Various methods exist worldwide for the treatment of patients with a Robin sequence, many involve major surgery. Our treatment concept has been used for many years and has established itself successfully due to the comparatively low stress for both, the child and its parents. It consists of three main pillars:

Treatment pillar	Responsible specialists
1. Tübingen palate plate	Orthodontics, neonatology
2. Drinking and swallowing training	Speech therapy, pediatric nursing
3. Surgical closure of the cleft palate	Oral and maxillofacial surgery

1. Tübingen Palatal Plate (TPP)

Various methods are available worldwide to correct the narrowing of the upper airway in affected patients. These range from positioning the infant in a prone position, inserting a breathing tube into the throat (nasopharyngeal tube) to various surgical procedures. Examples of surgical procedures include suturing the tongue to the lower lip (glossopexy), pulling the lower jaw forward using wire loops and a small weight (wire extension), gradual bone expansion of the lower jaw (mandibular distraction osteogenesis) and a tracheotomy.

Since most methods require surgical intervention and have other risks and disadvantages, a less stressful treatment concept for these children and their parents was developed in Tübingen in collaboration with pediatricians, orthodontists and oral surgeons: the Tübingen palatal plate.

This is a special, slightly longer palatal plate (similar to those used in children with a “simple” cleft palate) with an integrated spur or extension, which is inserted into the mouth and brings the tongue forward.

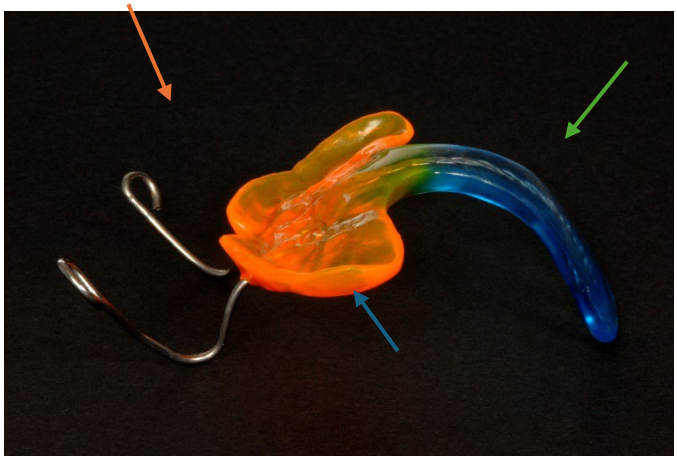

Illustration TPP; green arrow = spur, orange arrow = holding spines, blue arrow = palate-covering part

The Tübingen palatal plate prevents the tongue from falling back, eliminates any constriction in the throat and helps to stimulate growth of the lower jaw. By moving the tongue forward from the nasal into the oral cavity, the Tübingen palatal plate supports not only breathing but also the child's food intake, sound formation and therefore speech development as well as a normal swallowing pattern. It also has a positive effect on the function of the ear trumpets (tubes) and the ventilation of the middle ear.

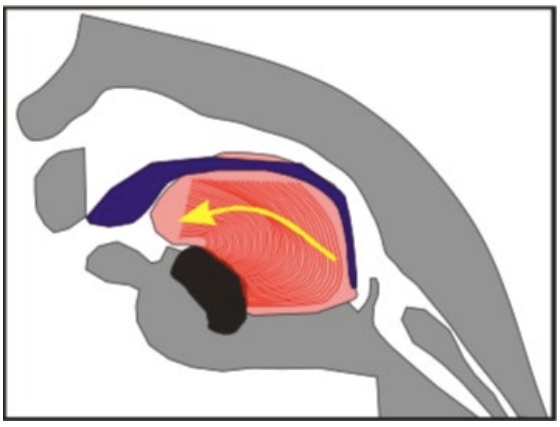

Illustration 3: Effect of the TPP

In addition to the important functional separation of the oral and nasal cavities, early plate fitting also brings the jaw segments closer together and thus narrows the cleft in patients with an additional cleft palate. This also improves the conditions for the subsequent surgical closure of the cleft.

There are no reports of negative effects of the Tübingen palatal plate.

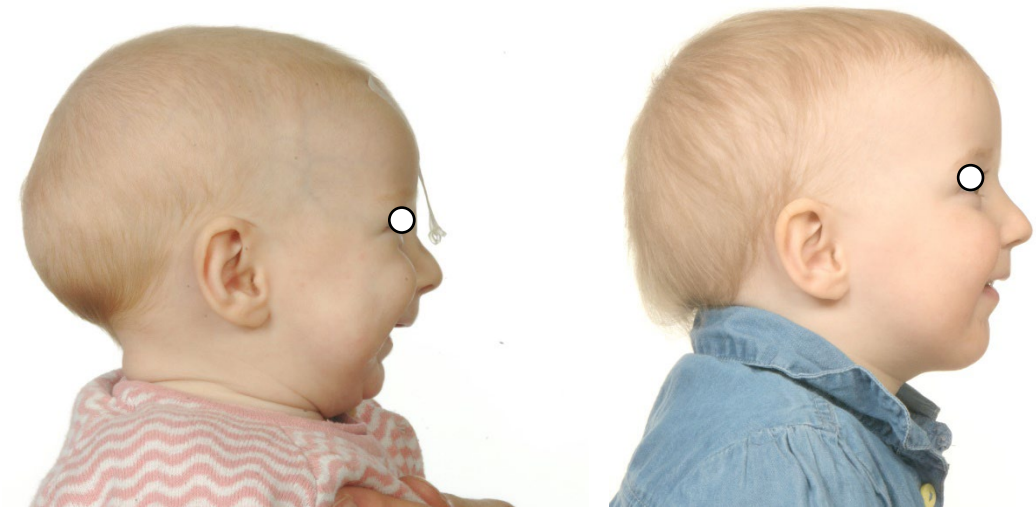

Illustration 4: Patient with RS before TPP therapy at the age of 2 M and after TPP therapy at the age of 1 J 3 M

2. Drinking and Swallowing Training

The second important pillar of the treatment concept is to help babies to learn how to suck and swallow through a combination of experienced nursing care and speech therapy, the latter based on Castillo Morales® and Padovan. Our long-term aim is to enable patients to adopt and maintain a physiological tongue position even without the Tübingen palatal plate. This helps to stimulate the growth of the lower jaw and normalizes food intake. The child is placed on the practitioner's lap, who slowly works their way towards the mouth during the exercises.

Parents are taught the basic exercises from the outset so that they can carry out the training independently at home with their children at least once a day after discharge from hospital. The child should be awake and each exercise should be repeated around four to six times. Parents are also shown special exercises that should be performed prior to each meal.

3. Surgical Closure of the Cleft Palate

If the child with RS has a cleft palate, this should be surgically closed after finishing TTP therapy. The closure of the palate separates the nasal cavity from the pharynx, which is important for food intake and speech development, among other things.

During the operation, the function of the soft palate (velum) is restored by reuniting the cleft palate muscles and closing the mucous membrane towards the nasopharynx and the oral cavity. If the cleft palate extends further forward, i.e. if the hard palate is also affected, this is also closed in the same procedure by moving the mucous membrane.

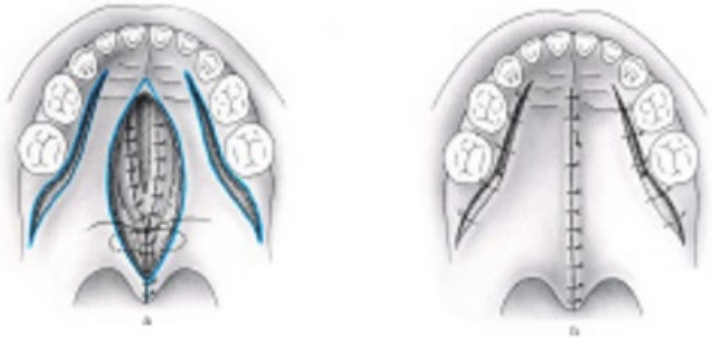

Illustration 5: Schematic representation of plastic cleft palate closure (from Horch 2007)

The Treatment Process

What Patients and Affected Families can expect from us.

1. Prenatal Diagnostics

Ideally, a suspected diagnosis of Robin sequence should be made before birth. This is done using prenetal ultrasound in which the lower jaw is found to be too small and shifted backwards. In addition to the purely subjective assessment of the lower jaw, angle measurements, for example between the lower jaw and nose or forehead, can also be used. More often, however, the diagnosis of Pierre Robin sequence is only made at birth.

The advantage of prenatal diagnostics is that families can find out about RS and its various treatment options in advance and can choose a delivery hospital that specializes in these diseases and therefore has experience with affected patients.

Prenatal diagnosis of cleft palate, which is often associated with a Robin sequence, is unfortunately still a major challenge.

2. Inpatient Stay

Immediately after birth, babies with RS are initially cared for by us in the neonatal department of the children's hospital. After admission, the severity of the respiratory disorder is assessed by recording the baby’s breathing signals during sleep (polysomnography). Obstructive apneas, as described above, cannot be reliably detected with conventional monitoring in the hospital or through a home monitor, so that a polysomnography is necessary to assess the sleep disorder.

The examination is completely painless, is carried out exclusively with adhesive sensors and includes the recording of chest, abdominal, nasal and mouth breathing, as well as the heart rate, blood oxygen and carbon dioxide levels and sleep movements. The child is connected to a recording device overnight and should sleep on its back during this time if possible. The recording is then analyzed and the extent of the breathing disorder determined.

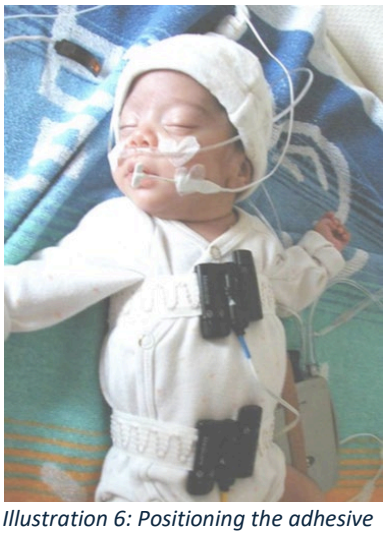

Illustration 6: Positioning the adhesive sensors for the sleep laboratory

In addition, the patient's upper jaw is scanned promptly to produce and fit the Tübingen palatal plate. This is carried out using a low-risk 3D intraoral scanner, whereby the scan only takes a few seconds and can be performed without sedation or anesthesia.

At the time of initial insertion of the Tübingen palatal plate, the extent of the obstruction of the airway is visualized using endoscopy, and other causes of a respiratory disorder are ruled out at the same time. The length and shape of the spur of the palatal plate in relation to the base of the tongue and epiglottis is also adjusted individually for each child during the examination. The endoscopy only takes a few minutes and can be performed without sedation or anesthesia, only under local anesthesia.

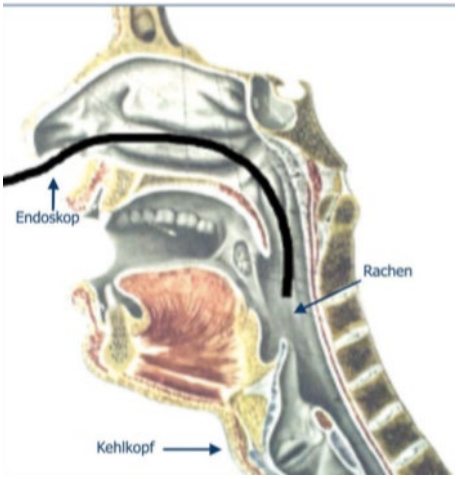

Illustration 7: Endoscopy of the pharynx

Illustration 8: Insertion of a TPP with bonding cream

As a next step, the Tübingen palatal plate is shaped into its final form, reinforced by a metal wire, fitted with additional retaining brackets and then inserted with the help of standard denture adhesive cream.

If the plate fits well and the baby accepts it, its effectiveness is checked after at least 48 hours by means of a new polysomnography, where the need for further improvement of the Tübingen palatal plate is verified.

3. Discharge from the Initial Hospital Stay

Parents are fully integrated into the treatment at an early stage so that they can continue their baby's treatment at home. As soon as parents have mastered the daily, independent changing of the Tübingen palatal plate, feeding their child and performing the essential elements of functional physio- and speech therapy, discharge is possible. The plate should always remain in the patient's mouth and only be removed once daily for cleaning and refixing. Cleaning is carried out with a toothbrush and water and, if necessary, with a little fluoride-free toothpaste. In the mouth, adhesive cream and food residues should be carefully removed with a compresse. In this context, the mucous membrane and palate can be checked for potential pressure marks.

The average duration of therapy for fitting the Tübingen palatal plate with spur, achieving independent feeding without a feeding tube and teaching parents how to change the plate is around 3 weeks. The duration of treatment is usually somewhat shorter if treatment is started in the newborn period. Longer stays may be necessary for children with an additional syndromic disease.

A home monitor (pulse oximeter) is also prescribed in the first year of life to monitor the oxygen saturation.

The overall duration of the TPP treatment is determined individually over the course of the treatment and is usually 6-9 months. As the child grows, 1-2 new adjustments of the Tübingen palatal plate are usually necessary, according to our experience every 3-4 months or after a weight gain of approx. 3 kg. An intraoral scan is taken again and a new plate is fabricated in the same way. If the teeth break through, it is not necessary to make a new plate; it is sufficient to remove material from the relevant area. For the duration of the palatal plate therapy, check-ups are carried out every six to eight weeks.

4. Surgical Closure of the Cleft Palate

In the course of the check-ups, the time for cleft palate surgery is determined together with the parents. The operation is usually performed around the first birthday. In order to give the child time to grow and the lower jaw time to develop, the operation is generally not performed before the child is eight months old. Before this, we carry out another sleep laboratory examination to make sure that the baby is still breathing well after completing the Tübingen palatal plate therapy.

For the night of the operation, a bed is reserved in the children's intensive care unit for better monitoring of breathing, although this is not always required. On the fifth day after the operation, discharge from inpatient treatment is possible if the child is drinking well.

As the entire palatal surface has changed after the operation, the Tübingen palatal plate can no longer be used and is then no longer necessary.

5. Regular Checks

Our interdisciplinary collaboration at the center ensures that those affected continue to receive care until reaching adulthood. Affected children are monitored several times in the first few years and at least once a year subsequently, depending on their development. Speech development, upper and lower jaw growth, hearing and dental development are checked.

If necessary, you will be offered orthodontic follow-up treatment as part of these check-ups. This is usually carried out as an early orthodontic treatment from the age of 4 years or as a main orthodontic treatment starting around the age of 8-9 years.

As the psychological strain on parents can be great, our psychosocial service is available to parents with advice and support - even after discharge from hospital.

The Tübingen Team

Figure 9: from left to right: PD Dr. Cornelia Wiechers, Dr. Kathrin Böckmann, Lea Longerich, Dr. Katharina Peters, Prof. Dr. Michael Krimmel, Prof. Dr. Dr. Bernd Lethaus, Prof. Dr. Christian Poets, Prof. Dr. Bernd Koos, Dr. Kathrin Heise, Margret Fritz, Dr. Christian Maiwald, Dr. Karen Kreutzer

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- Neonatology and interdisciplinary sleep medicine
  Prof. Dr. Christian Poets (Medical Director)
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Parental consent has been obtained for all images of patients.