

22q11 deletion syndrome and speech and language development in the pre-school years

Information for families

North Thames Regional Cleft Lip and Palate Service
Great Ormond Street Hospital / St Andrew's Centre

This information sheet explains about speech and language development in children with 22q11 deletion syndrome, focussing on the years before they start school. It also gives information on feeding and swallowing. The final section suggests ways that you, as a parent, can help your child's speech and language development.

What is 22q11 deletion?

22q11 deletion syndrome is a type of chromosome disorder that is found in many seemingly unrelated conditions or syndromes (collection of symptoms often seen together). Human beings have about 30 to 40,000 different genes, each of which has a function in making an individual person. The genes are arranged in pairs (one of the pair from each parent) on 23 chromosomes. Each chromosome has a long arm (referred to as q) and a short arm (p). In 22q11 deletion, a tiny part of the long arm of one of the two copies of chromosome 22 is missing at position 11.

It is diagnosed by a special blood test. You might hear the condition being called by other names as well, such as 22q11.2 micro deletion syndrome, velocardiofacial syndrome, Shprintzen syndrome or di George syndrome.

Children with 22q11 deletion syndrome can be affected in a variety of ways. They may have heart problems, reduced immunity, feeding difficulties, palate problems and difficulties with speech, language, hearing and general learning. It is important to remember that every child is different. Always talk to the doctors and healthcare professionals who are looking after your child if you have any worries.

Feeding difficulties

Approximately 40 per cent of children with 22q11 deletion syndrome have some feeding difficulties. These usually occur during the first year of life, and often resolve as the child grows and develops, but in some cases, they may last throughout childhood. Problems are often related to medical difficulties such as heart or digestion problems, but not always. Early difficulties include not being able to suck well from the teat or nipple, smaller volumes of milk taken at each feed, and refusal of the bottle/breast and/or food. Some young infants are slower to develop chewing skills, and can take longer to progress from early baby foods. They may store chewy foods in the side of their mouth. Your child may also show a strong preference for certain foods or textures.

Swallowing difficulties

A small proportion of children may experience swallowing difficulties, known as dysphagia. This may show itself as coughing or distress when eating and drinking, combined with frequent unexplained chest infections.

Language skills

Children with 22q11 deletion syndrome are often late talkers. You may find that your child babbles less than other children of a similar age. At around the age of 18 months, there is often both slow development of the understanding of language (known as receptive language) and a more significant delay in starting to talk (known as expressive language).

Often at this early stage, when language skills may be very limited, children benefit from using sign language. In the UK, we use a system called Makaton® sign language, which is based on simple single words where the adult always says the word at the same time as using the sign to encourage

language development. Children with 22q11 deletion syndrome often take to signing very well and will even make up their own signs, dropping the signs once they are able to say the word.

Children with 22q11 deletion syndrome benefit from assessment and monitoring of their speech and language by a speech and language therapist. The therapist can then plan with you how to best help your child's development in these areas. Often your child will be under the care of speech and language therapists in both the hospital and your local clinic or child development centre. At the hospital, we will be concentrating on specific areas that relate to the diagnosis of 22q11 deletion syndrome, in particular your child's speech and whether there is any air coming down his or her nose in speech (see below about nasal speech). Your local speech and language therapist will concentrate on your child's overall speech and language development and usually will be working alongside nursery and other education staff.

Speech skills

The development of speech sounds (how a child pronounces words) is often particularly delayed in children with 22q11 deletion syndrome. They also have a tendency to disordered speech development (making unusual sounds). For example, they may miss out sounds both at the beginning and ends of words, or use lots of hard unusual sounds made down in the throat. They may use very few or no sounds made in the mouth, such as /b d g/.

If your child is showing these patterns of speech, he or she will need to be seen by a speech and language therapist. Your child may also need a referral to a regional cleft team to assess whether these difficulties are related to how the palate and back and side walls of the throat are working (see nasal speech section below).

Children with 22q11 deletion syndrome can also have difficulties with how they coordinate their tongue, teeth, lips, palate and air from their lungs to produce speech. These problems mean that

children can get sounds muddled up, in the wrong place, or miss lots of sounds out. Your speech and language therapist will be able to advise you on this.

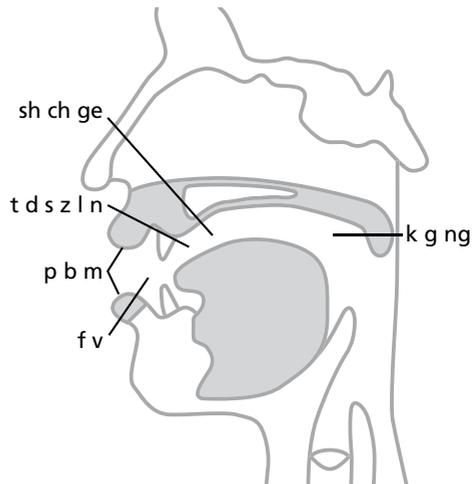
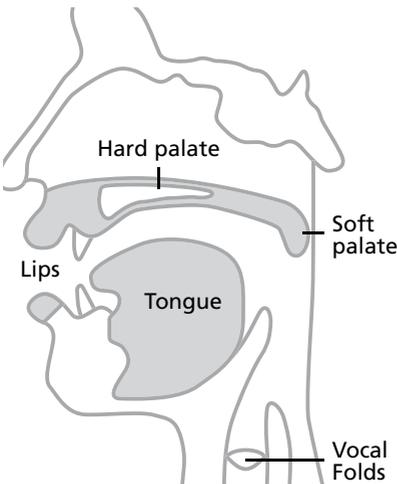
Nasal speech

We produce speech by air coming from the lungs, through the vocal folds (voice box) and out of the mouth. We shape the sound with our tongue, lips and teeth to say words and sounds. The diagrams below show the main parts of the speech mechanism, the consonant sounds in English and where these are made.

The soft palate plays an important role in helping us talk, as it moves to close off the nose from the

mouth during speech. Most sounds we use in the English language are made with the palate raised (with the nose cavity closed off) resulting in 'oral' sounds (p, b, t, d, k, g, f, v, s, z, sh, ch, ge). In English, there are three sounds (m, n and ng) where the palate is lowered (the connection between the mouth and the nose is open) resulting in 'nasal' sounds.

Problems with nasal speech are particularly common in children with 22q11 deletion syndrome. The causes of these problems with the soft palate closing off against the back and side walls of the throat may be due to many factors and so specialist assessment by a regional cleft team is needed.



When children have nasal speech, the overall sound of the voice is nasal. Nasal air emission (where air can be heard escaping through the nose during speech) or nasal turbulence (a more noisy form of nasal air emission), may also be present. It is important to find out why. Your child may need to have two simple investigations – a videofluoroscopy (x-ray while speaking) and a nasendoscopy (a small camera passed through one side of the nose). These tests show how the soft palate and back and sides of the throat work during speech. Please see a copy of our *Investigations of nasal speech* information sheet for further information. The speech and language therapists may also make speech/audio/video recordings and sometimes measure the sound of your child's voice with a computer. These investigations will help your child's surgeon and speech and language therapists decide on the best treatment to solve this problem, which can include surgery.

Starting school

If your child is experiencing problems with speech and language and other areas of his or her development, it is likely that your child will be under the care of your local child development team. They may recommend that he or she be referred to your local educational authority for an assessment. This will help make sure that any extra assistance your child needs when starting school is in place. If you are concerned about your child starting school, do speak to your local paediatrician or the speech and language therapists working with your child.

Activities to help your child's early language development

Listening to sounds

Point out new sounds to your child and always show what makes the sound – for example, “that’s the doorbell ringing”. Introduce your child to ‘symbolic’ sounds, which are sounds which carry a meaning of something, like “brrmm brrmm” when you see or hear a car, and animal noises like “meow” when he or she sees a real cat or a picture of one.

Taking turns

Play games that encourage taking turns, such as ‘peek a boo’ with a piece of cloth or rolling a ball to each other. Encourage your child to look at you in these types of games. Make a game of giving and taking things as your child starts to understand your requests – for example, “give me the car”, “give me the teddy”, then give these back to your child saying, “here’s the car!”.

Play

Give your child things to feel and look at, such as a baby mirror or soft cuddly toys, and things to hold, shake and bang – for example, bells and bricks. A wooden spoon and saucepan lid can make a brilliant drum! You can also make shakers from washed small plastic drinks bottles filled with dried rice, pasta or lentils – just make sure you secure the lids carefully. Encourage your child to follow toys with his eyes. For example, you can blow bubbles or balloons in front of your child or use finger puppets. You can also play hiding games with your child – let a piece of tissue or scarf fall over your head or cover a toy and encourage your child to pull it off. Play ‘pretend’ games with your child – for example, have pretend cups of tea or bath a doll together. You could also have fun feeding teddies or dolls and putting them to bed. Signing and talking while doing this will help your child learn new words which are very relevant to everyday activities.

Nursery rhymes and songs

Listen to nursery rhymes or songs with your child or sing your child's favourite nursery rhymes. Carry out the actions as you sing the words – for example in the song 'The wheels on the bus go round and round'. When the songs and the words become familiar to your child, encourage him or her to fill in the sounds, actions or words which come at the end of the line – for example, 'Heads and shoulders, knees and ____' You can also watch your child's favourite DVD with him or her. See if your child can join in with the actions and words of the nursery rhymes and songs. Please do remember to just let your child watch television for short periods of time only. Do switch the television off then so that you keep background noise and distractions to a minimum.

Looking at books

Look at books with your child. As you look through the book, name and point to a familiar picture of an object in the book, for example, "I can see the duck" and then ask your child to point to by saying

"can you see the ____?" In the early stages your child will not want to look at a page for long. Go at his or her speed and gently point things out in the book as you turn pages.

Everyday routines

Your everyday routines are a great way to help your child learn new words. You can begin to talk about parts of the body as you give your child a bath, with for example "Let's wash your tummy". You can also have great fun at bath times taking turns putting bubbles on each other's nose or hair! You can also use nappy changing and getting dressed to talk to your child stressing this everyday vocabulary and introducing new words too, like cream, shoes and socks.

A visit to the playground can be another great opportunity to use new words like flowers, sand and swing. You can also introduce words such as 'up', 'down', for example, "You are coming down the slide!"

Learning new words

Help your child learn new words by emphasising these, saying “Isn’t your ice-cream cold. . .” or “do you want more spaghetti?” Try to find opportunities to use the new word over and over so that your child becomes familiar with it. It also helps if the word is about something that interests your child.

Learning to categorise words that go together

As your child is playing, you can say the words for the toys he or she is playing with, for example car, ball, teddy – and then say the word ‘toys’. Another good opportunity is mealtimes where you can use the word food to talk about all the different things we eat. In this way your child can learn about categories of words.

If your speech and language therapist has been showing you how to use Makaton® signs with your child, do remember to both say and sign the new words. This will help your child learn these.

Give your child as many opportunities as you can to use the words and signs he or she has learnt. For example, keep his or her favourite toy out of reach but where he or she can see it. See if your child will ‘ask’ or sign for it before you fetch it. Remember, it is important that your child does not get frustrated and is rewarded for trying to communicate, so if he or she cannot speak or sign clearly yet, accept his or her particular way of asking, which may be looking, pointing, reaching dragging you towards the toy or making a noise. You can then say and sign the word back to him or her clearly, for example “Oh, you want teddy” and then praise your child for their efforts.

General activities to help speech development

Getting ready to play

You can play sound games at any time. It is generally best to try to cut down on background noise so that your child can hear your voice. Also try to sit facing your child so that he or she can see your face and mouth clearly.

Babble with your baby

Listen to the sounds your child makes and repeat these back to him or her. This will allow your child to hear and see the sounds he or she is making and let you tune into his or her speech sounds. Let your child see and feel your mouth moving either with his or her hands or on his or her skin. For example, blow raspberries and let your child touch your lips to feel the vibration. Do remember, if your child is making lots of sounds in his or her throat, avoid repeating these back. Instead repeat a mouth noise back.

Starting talking

Encourage your child to make sounds. Children with 22q11 deletion syndrome at this early age usually won't be able to copy sounds perfectly. Accept what your child can say and repeat the word back clearly yourself. For example, if your child says "a" for car, you can say, "Yes, it's a car".

Teach new sounds

Encourage early speech sounds, particularly gentle lip sounds such as "muh, buh, puh". Use words like "peep-o", "pop" and "mummy" or imitate animal sounds, for example, "moo", "baa" and "meow", or make up nonsense strings of sounds, for example "mumumum" or "boobooboo". If you make the sounds, your child will want to join in. Pop bubbles while saying "pop, pop, pop" and play hiding games, saying "peep-o", bounce a ball saying "buh, buh, buh".

Further information

For further information regarding your child's speech and language development, please contact your local speech and language therapist or the speech and language therapist attached to your child's regional cleft team, where appropriate.

Max Appeal

Tel: 0800 389 1049 (free 24 hour answer phone service)

Website: www.maxappeal.org.uk

VCFS Educational Foundation (US)

Website: www.vcfsef.org

International 22q11 Deletion Syndrome Foundation (US)

Website: www.22q.org

Notes



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