



Great Ormond Street Hospital for Children NHS Trust: Information for Families

Your child with 22q11 deletion: what you can expect from GOSH

Throughout your son or daughter's childhood, your family doctor (GP), local paediatrician and child development centre are likely to remain the main health service providers for his or her needs. Staff at GOSH will of course be available for advice and discussion if needed. This information sheet gives you a guide to the tests and investigations your child will need throughout childhood to make sure that he or she is in the best health possible. It is intended as a quick guide for you and your health care team.

Basic investigations

- Everyone should have these after the diagnosis of 22q11 deletion has been confirmed by the FISH test:
 - Heart assessment, including echocardiogram (Echo)
 - Renal ultrasound
 - Immune functions tests, including lymphocyte count and immunoglobulins IgG, IgA and IgM
 - Blood tests, including full blood count and blood calcium level

Immunisations and infections

- Confirm with your paediatrician whether 'live' vaccines can be given.
- Antibody level should be checked six weeks after the last '5 in 1' vaccination and the MMR. If the levels are low, another dose is advised to boost resistance.
- BCG (vaccine against TB) is a live vaccine so should only be given if specifically indicated by a specialist.
- Infections may be frequent and benefit from specialist advice.
- If your child is in contact with chicken pox, tell your doctor immediately as treatment may be needed.

Investigations essential during growth

- Height and weight check every one to two years by GP. Plot the results in your child health record ('red book').
- Thyroid function tests are usually done if growth is slowing or your child is small. Tests for growth hormone are occasionally needed.
- Calcium levels in the blood should be checked if movements are 'jittery' or your child complains of limb or back



pains. Calcium levels should be checked every year up to the age of five and at puberty when growth is rapid from the age of 10 to 15 years.

Feeding

- Bottle-feeding may be slow, there may be a delay in moving to more solid food or he or she may be quite selective about what food is preferred.
- Constipation can be troublesome.
- Swallowing may be disorganised. Symptoms to watch out for include food or drink coming down the nose, frequent chestiness, coughing/choking or colour changes during feeding and/or constant wheeziness. Your paediatrician or speech and language therapist can advise whether specialist support is needed.

Dental

- Checks from infancy should be regular, especially if a heart problem is present.
- Teeth may be more likely to develop holes so prevent this by regular brushing with children's toothpaste containing fluoride.
- Always tell your dentist about a heart condition if one is present.

Speech and language difficulties

- These are common so assessment and monitoring by a speech and language therapist is usually required.
- Your child may have some difficulties understanding language so may take longer to learn to talk. Language skills may need to be monitored throughout school years. The use of natural gesture and signing alongside speech is recommended to help development of communication, particularly in the preschool years.
- Speech may be difficult to understand due to problems pronouncing words and 'nasal sounding' speech. Specialised investigations of the palate and back of the throat may then be advised. Depending on findings and progress, surgery may be recommended.
- Your speech and language therapist can advise on assessment and treatment for your child's difficulties.

Learning difficulties

- Developmental progress and learning ability are often affected. If you or a teacher is concerned, it is advisable to start looking for help early. There are several stages in getting educational support, so it is a good idea to start the process even before starting primary school.
- At four to five years old, an assessment by a clinical psychologist may be arranged if we think that it is indicated.



- The change from primary to secondary school is another key stage that might need careful assessment. It is best to start with your child's school's special educational needs coordinator (SENCo) by the age of 10 years.
- If difficulties arise, you can contact our clinic coordinator who will pass your concerns on to a member of the team at GOSH.

Behaviour

- This can be challenging, difficult or withdrawn at any stage throughout childhood. Rapid changes in mood, difficulty in concentration and anxiety are common. These can be discussed at our clinic after your child has reached the age of five years or you can ask your family doctor or health visitor for a referral to the local child and family counselling service.

Pains affecting legs and wrists

- These are common and have many causes, mainly relating to lax ligaments.
- Swelling and stiffness of the joints with pain might be a kind of arthritis and needs early assessment by a paediatrician.

Mental health

- These problems might surface during young adult life. Indications for discussion with your family doctor

include 'hearing' people talking about him or her, severe social withdrawal, early morning waking, obsessive thoughts and/or depression.

Genetic counselling

- This may become important when your child reaches the stage where they will want the information and be able to understand it. This usually happens around the age of 15 to 20. Remember that you can also ask for referral to a genetic counsellor if you are considering pregnancy and want up-to-date advice.

Other problems

- Other medical problems might be present or emerge at any point during your child's life. Always discuss these with your family doctor, reminding him or her of your child's diagnosis of 22q11 deletion. If they wish, they can contact the cleft coordinator who will arrange a reply from the most suitable member of the team at GOSH.

Follow up

- We hold our 22q11 deletion clinics for children aged two years or older and repeat them at the age of four or five, 10 and 15 years old. These times are usually when decisions about education are necessary and discussions about behaviour or medical progress can be helpful.



Further information

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

For further information, please contact our clinic coordinator on 020 7405 9200 ext 7922 Monday to Friday 9am to 5pm

Useful numbers

GOSH switchboard 020 7405 9200

Speech and Language Therapy Department 020 7813 8110

Cleft Lip and Palate secretaries 020 7813 8242

Cleft Service fax 020 7813 8279

Notes

Compiled by the 22q11 Deletion Syndrome team in collaboration with the Child and Family Information Group

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www.goshfamilies.nhs.uk

www.childrenfirst.nhs.uk