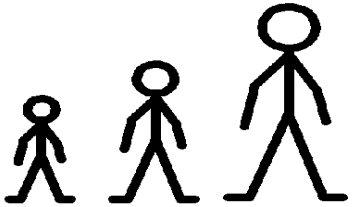
Personal Health Record



22q11 Deletion Syndrome

Central Manchester University Hospitals NHS
NHS Foundation Trust

Contents

22q11 Personal Health Record

You have been given this booklet because you/your child has a Chromosome 22q11 deletion syndrome. Some other names associated with this condition are, DiGeorge Syndrome (DGS) and Velocardiofacial syndrome (22q11). In this booklet we will use the term '22q11 deletion' to refer to all of these conditions. People with a 22q11 deletion may see a number of doctors and specialists, and this booklet will help you and your health care team ensure you receive the correct care and appropriate check ups. Please complete pages 4 and 20 yourself and bring the booklet with you whenever you visit the genetics clinic or any of the following and ask them to complete the relevant pages:

- Your Paediatrician/Community Paediatrician *(ask them to complete pages 13, 14, 15, 16, 17, 18 & 19)*
- Your GP *(ask them to complete page 16)*
- Hospital Outpatients Clinic or Emergency Departments *(ask them to complete pages 5 or 6, 16, 20 & 21)*
- Your Dentist *(ask them to complete page 16)*
- Your Optician *(ask them to complete page 16)*
- Any other health care appointment *(ask them to complete page 16)*

You may also wish to use the plastic pockets in the front and back of this folder to store appointment letters, letters from your GP and hospital doctors, or anything else related to your 22q11 that you want to keep safe.

Spare pages for this booklet are available from the Regional Genetics Service.

Important Note: This record does not replace your hospital, GP or other medical records which will continue to exist alongside this one.

This is the Personal Health Record of:

Name		**GM No.**	
Address		**Date of Birth**	
		Telephone	☎

Useful Contacts:

Main 22q11 Doctor	NAME	☎
GP	NAME	☎
Cleft Palate Team	NAME	☎
Dentist	NAME	☎
School (if applicable)	NAME	☎
Health Visitor	NAME	☎

22q11CLINIC DETAILS

Genetics Service should attach a printed sticker here with clinic address and contact details.

Specialist Doctor Details

Sheet Number

People with 22q11 deletion are often looked after by more than one specialist, and this page is a record of all the doctors who are involved in caring for you/your child. If you visit a specialist doctor who isn't listed, they should add their details below.

Name	Title/Specialty	Address/Phone Number
		☎
		☎
		☎
		☎

Specialist Doctor Details….continued

Sheet Number

People with 22q11 are often looked after by more than one specialist, and this page is a record of all the doctors who are involved in caring for you. If you visit a specialist doctor who isn't listed, they should add their details below.

Name	Title/Specialty	Address/Phone Number
		☎
		☎
		☎
		☎

Professionals Involved in Your Care

People with 22q11 deletion are often looked after by a team of specialists. This page is a brief description of the various people you might visit and the role they play in your care.

Paediatrician/Community Paediatrician
The paediatrician/Community paediatrician looks after the growth, general health and development of babies and children.

Cardiologist
A cardiologist looks after the health of your heart.

Cleft Palate Team
If you or your child have a cleft palate or nasal speech this will be looked after by a team that includes: a Plastic Surgeon who is responsible for surgery to repair the palate, an Orthodontist who will work with the plastic surgeon to align the jaw and teeth, an audiologist who specialises in hearing loss and correcting hearing problems, a dental hygiene practioner will advise on the care of teeth and gums, a speech therapist who monitors language development and gives advice to improve the quality of speech, and a specialist health visitor who advises on feeding and liaises between you and the other members of the cleft palate team.

Community Speech & Language Therapist
Provides speech and language therapy locally.

Clinical Geneticist
Has a role in identification in the cause of birth defects and will discuss this with you. Provides advice to help with the management of rarer disorders and discusses the implications for future pregnancies and the extended family.

Genetic Counsellor
Works with the clinical geneticist to provide support and advice to families.

Educational Specialists
An educational psychologist will assess your child if there are any concerns about development or behaviour. Other education specialists work with the local education authority to ensure that educational needs are met.

Endocrinologist
Investigates and treats hormonal problems. For example if you have a thyroid problem, your energy levels and growth may be affected.

ENT (Otolaryngologist)
Will see your child if there is a need for surgical treatment of the ear, nose or throat, e.g. to insert grommets to correct glue ears.

Clinical Psychologist
May be involved to manage behavioural feeding difficulties, difficult behaviour and if a more comprehensive assessment of development is needed. Some children may be referred to a psychiatrist.

GP (Primary Care) Services
Your GP is responsible for health matters on a day to day basis. GPs work with Health Visitors to support young families and monitor development and immunisation.

Immunologist
Your immune system protects your body against disease. An immunologist monitors your immune system and advises on treatment if your body is not fighting infections as effectively as usual.

Ophthalmologist
Specialises in the diagnosis and treatment of eye conditions.

Social Worker
Liaises with the other professionals involved in your care and can provide help with housing and benefits.

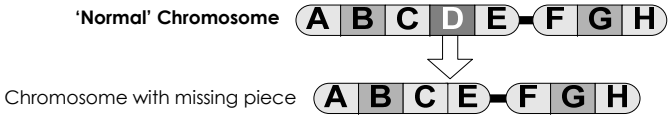
Information for You and Your Family (part 1)

What is 22q11 Deletion?
22q11 deletion is a genetic disorder. You may also hear it called DiGeorge Syndrome (DGS) or Velocardiofacial Syndrome (VCFS). A syndrome is a combination of symptoms which grouped together form a recognisable pattern.

The way in which a 22q11 deletion affects someone varies from person to person. Some people will have no noticeable symptoms of ill health, while others will have multiple problems from birth. Because 22q11 deletion is associated with a range of health problems, it is recommended that children with this condition have regular checkups.

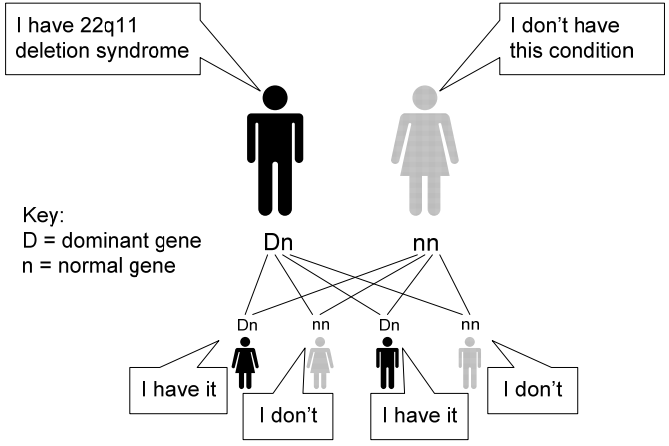
What is a 22q11 Deletion?
It is an abnormality of one of your chromosomes. Chromosomes are small packages of genes which are responsible for individual characteristics such as eye and hair colour. People with 22q11 deletion have a tiny part of one of their chromosomes missing or 'deleted'. This small portion contains genes which are important for development of the heart and palate and for general development.

Chromosome Deletion



The majority of people who have a 22q11 deletion are the first person in their family to be affected. A small number may have inherited a 22q11 deletion from one of their parents. This condition is 'dominant' which means that if you have a 22q11 deletion, there is a 1 in 2 chance of passing this on to each child.

How 22q11 syndrome can be passed on from a parent to a child



Information for You and Your Family (part 2)

What are the Features of 22q11 Deletion?

22q11 deletion is a complicated condition affecting many different parts of the body. Some of the most common features include:

- Palate Abnormalities — cleft palate or more subtle abnormalities of the roof of the mouth (palate), and throat (pharynx) leading to problems with feeding and speech.
- Heart Defects— usually present at birth (congenital). These vary from a simple hole in the heart, to more complex heart problems which may require surgery.
- Learning Disability—some children will walk and talk later than usual. Language delay in particular is common
- Hypocalcaemia— low levels of calcium in the blood caused by hormone imbalance which can cause painful muscle spasms, and seizures.
- Immune deficiency— prone to recurrent infections, particularly of the chest and ears in children.
- Behavioural Difficulties— ADD, ADHD, psychiatric problems may be a feature in some children and adults.
- Kidney problems— differences in the way that the kidneys develop may lead to urinary tract infections but most don't affect kidney function.
- Short Stature—some people with a 22q11 deletion are short as children, many of them catch up after puberty and attain a normal adult height.
- Feeding Difficulties - these may also be worsened by heart problems, reflex and problems with the muscles of the throat. Some feeding problems may be behavioural.

Additional health problems include:

- Hearing and throat problems
- Hormone imbalance
- Problems with bones and back
- Constipation
- Leg Pains
- Squints and rarer problems with vision

What are the Physical Features?

People with 22q11 may look similar, with a small mouth and chin, a broad bridge to the nose, sometimes round and prominent ears. Facial features change with age and the nose may become slightly more prominent in adulthood. Sometimes there may be dark circles under the eyes, these are nothing to worry about. Other physical features include slender, tapered fingers.

Management of 22q11 Deletion

Feeding
The majority of babies with 22q11 deletion will have some difficulties feeding due to problems with the muscles at the back of the throat. Referral to a feeding specialist is often needed. Special feeding equipment is available if needed.

Immune System
Children with 22q11 deletion are more prone to infections because they do not have as many T Cells which fight infection. Most of these will be mild but occasionally children with more severe infections will need more specific treatments. All children should have their immune system checked and those with a problem should be referred to an immunologist. Care should be taken with immunisations and live vaccines should be avoided in children with immune system problems.

Leg Pains
These are frequent in children with a 22q11 deletion and treated best by orthotic appliances in the shoes

Calcium Deficiency
About half of all people with 22q11 deletion will have a low calcium level. This may present with jitteriness or fits in young children or with muscle cramps or tingling of the mouth and fingers. Low calcium levels should be treated with calcium supplements under the supervision of a specialist who will monitor calcium levels and arrange regular kidney scans whilst on treatment. Milder symptoms can be treated by a milky drink at bedtime. Adolescence is a common time for calcium problems.

Development
Some people with 22q11 deletion will have entirely normal development, but often milestones (eg walking) are delayed and early intervention is needed. IQ usually falls within the normal range but it is important for development to be monitored on a regular basis. Expressive speech is particularly affected and at school children may have difficulties with comprehension and problem solving. Most children will attend mainstream school but will need extra help in the classroom.

Social and Behavioural Issues
Social skills are often underdeveloped making it difficult to make friends. Small size and unclear speech my contribute to low self esteem. Children are more at ease in familiar situations and with people they know well. Poor memory and attention span may be a problem but medication for this should be avoided since there may be adverse reactions.

Palate
Surgery is carried out to correct defects in the palate and throat and this together with speech therapy may improve swallowing and speech.

Heart
About two thirds of children are born with a heart defect. Many of these may not cause symptoms. All children with a 22q11 deletion should have a cardiology assessment including a heart scan. Surgery may be needed to correct severe heart abnormalities.

Information for Health Care Professionals

Diagnosis and Follow Up

22q11 deletion is a chromosomal condition occurring in approximately 1 in 4000 individuals. 10% to 15% of affected individuals inherit a 22q11 deletion from their parents, in the remainder it results from a spontaneous mutation. An affected parent has a 50% risk of passing on the gene to offspring.

The diagnosis of 22q11 deletion should be suspected on clinical grounds in individuals with multiple problems including cleft palate, velopharyngeal insufficiency, congenital heart defects, especially those involving the outflow tract, hypocalcaemia, learning disability and suggestive facial features. A variety of other anomalies may be present and on page 9. The diagnosis is confirmed by identification of a microdeletion of chromosome 22q11 detected on cytogenetic analysis using the FISH technique.

22q11 deletion is a variable condition. Signs and symptoms may vary over time and individuals may present in many different ways throughout their lifetime.

The 22q11 deletion is a multidisciplinary disorder requiring coordinated management by a number of professionals. In most cases, regular follow-up will be carried out by a general or community paediatrician with additional input from a variety of other hospital specialists.

The Role of the Regional Genetics Service

A clinical geneticist is often involved in the diagnosis of 22q11 deletion. They also have a role in ensuring patients receive appropriate follow up will be involved in genetic counselling of the families concerned.

Clinical Management guidelines and an information sheet for 22q11 deletion are available on request from the Regional Genetics Service or can be downloaded from our website **www.mangen.co.uk**

If you are still unsure about any aspect of caring for someone with 22q11 deletion, you can telephone the Regional Genetics Service or visit our website for specialist advice.

GENETIC CLINIC DETAILS

Genetics Service should attach a printed sticker here with clinic address and contact details.

Information for Education Professionals

How can 22q11 Deletion affect learning ability?
22q11 deletion syndrome is one of the more common genetic conditions that may have an impact on the way children and adults learn and perceive the world in which we live. The majority of children with a 22q11 deletion have an intelligence that falls within the normal range, however a significant proportion (>60%) experience difficulties that fall within the Specific Learning Difficulty spectrum. Severe learning difficulty in 22q11 deletion syndrome is unusual. Mean full-scale IQ scores on the Weschler scale range from 75 to 85.

A specific learning difficulty is identified when performance falls significantly below a child's general intellectual ability. For children with a 22q11 deletion, performance may fail to reflect their general intelligence. This can cause difficulties with comprehension, expressive speech, development of numerical concepts, and executive function (organisation).

What kind of problems could there be?
Children with a 22q11 deletion may learn to read satisfactorily and are good at spelling, but comprehension and expressive vocabulary can be poor. They are visual learners who prefer to 'watch and copy', rather than problem solve . Telling the time and 'money' work are difficult as are concepts of shape, colour and size, but arithmetical skills are usually good. Abstract reasoning may be problematic as children tend to think literally. Auditory processing may be impaired and they find it difficult to recall information without learned cues. Learning can also be affected by repeated absences from school due to hospital attendances.

Children with a 22q11 deletion may lack confidence and they are often easily frustrated and distractible . Sometimes they become obsessed with one topic and idea. They may display poor social skills in a peer group and have immature or inappropriate behaviour. Hyperactivity and attention deficit disorder are seen with increased frequency.

What steps should be taken?
Early identification and assessment of any difficulties is critical to ensure a child does not fall significantly behind their peers, and educators need to be aware of the condition. Small group or individual instruction is beneficial. The work environment should be quiet and complex tasks broken up into steps. Instructions should be clear and specific, using short sentences and repeating key words. Children benefit from a lot of repetition and routine and computer aided learning suits this group particularly well. Handouts or notes are helpful. Tests and examinations performance can be improved with help from word processors and allowing extra time. Staff should liaise with parents to reinforce skills at home. Participation in sports or musical activities should be encouraged. Strategies to support learning should be tailored to the individual child.

In itself, a 22q11 deletion is not a barrier to achievement and children with a 22q11 deletion can make substantial progress in response to targeted help. Whatever the child's difficulties there are ways of encouraging and supporting learning. However, it is easy to miss more subtle problems as those children may present in a very passive manner.

A Summary of How 22q11 Deletion Affects Me

Your main doctor will complete this page. It is a summary of how a 22q11 deletion affects you or your child. If other 22q11 deletion syndrome related symptoms arise between reviews, ask the Doctor you are seeing to add this to the summary. They will check that this page is up to date at your review appointments.

22q11 Related Features	**Date & Initials**	**Comment**	**22q11 Related Features**	**Date & Initials**	**Comment**
Heart			Kidney		
Palate			Education		
Vision			Behaviour		
Speech & Language			Leg Pains		
Calcium			Growth		
Immune			Hearing		
This summary is designed to help Health Professionals get a quick overview of how 22q11 deletion syndrome affects this person. Please only fill in positive findings . There is additional space on the next page to make more detailed notes, if necessary.					
Other Health Issues					
Medication Details					

Height and Weight Measurements (0 to 6 months)

Monitoring your child's height and weight is important. Your child's doctor will measure height, weight and head circumference at each visit and record them on the chart below. The standard growth charts are not entirely suitable for children with 22q11 as these children are usually small for their age.

	Birth		2 Months		4 Months		6 Months	
	Date:		Date:		Date:		Date:	
	cm	centile	cm	centile	cm	centile	cm	centile
Height								
Weight								
Head Circ								
Signature								
Comments								

Height and Weight Measurements (6 months and above)

Monitoring your child's height and weight is important. Your child's doctor will measure height, weight and head circumference at each visit and record them on the chart below. The standard growth charts are not entirely suitable for children with 22q11 as these children are usually small for their age.

Date	Age	Ht/Lth	Wt	H/C	Initials

Date	Age	Ht/Lth	Wt	H/C	Initials

Key Investigations Checklist

These important checks should all take place following diagnosis. This form should be used to record when these checks took place along with a brief note about the results.

Investigation	Date	Signature	Results	Comments
Heart Scan				
Kidney Scan				
Serum Calcium				
T Cell Studies				
Hearing				
Eye Tests				
Thyroid				
Dental Check				
Maternal Chromosome Studies				
Paternal Chromosome Studies				

Specialised Investigations Checklist

Sometimes, more specialist investigations are needed. This form should be used to record when these checks took place along with a brief note about the results.

Investigation	Date	Signature	Results	Comments
Developmental Assessment				
Video Fluoroscopy				
Spinal X Rays to check for vertebral/rib abnormalities				

Review Appointments With Your Paediatrician

Sheet Number

This page will be completed when you visit your paediatrician for a review. At this appointment, the doctor will carry out an examination and review any symptoms.

Date		**Doctor**	
Clinic Location		**Next Review Due**	

Doctors Comments

Date		**Doctor**	
Clinic Location		**Next Review Due**	

Doctors Comments

Review Appointments With Your Paediatrician

Sheet Number

This page will be completed when you visit your paediatrician for a review. At this appointment, the doctor will carry out an examination and review any symptoms.

Date		**Doctor**	
Clinic Location		**Next Review Due**	

Doctors Comments

Date		**Doctor**	
Clinic Location		**Next Review Due**	

Doctors Comments

Appointments With Other Specialists

Sheet Number

This page will be completed when you visit a specialist doctor.

Date		**Doctor**	
Clinic Location		**Next Review Due**	

Specialists Comments

Date		**Doctor**	
Clinic Location		**Next Review Due**	

Specialists Comments

Appointments With Other Specialists

Sheet Number

This page will be completed when you visit a specialist doctor.

Date		**Doctor**	
Clinic Location		**Next Review Due**	

Specialists Comments

Date		**Doctor**	
Clinic Location		**Next Review Due**	

Specialists Comments

My Notes

You can use this page for any other information you wish to remember or record. For example, use it to write down any questions you want to ask at your next appointment. Ask your Doctor for more sheets if you need them.

My Notes continued...

You can use this page for any other information you wish to remember or record. For example, use it to write down any questions you want to ask at your next appointment. Ask your Doctor for more sheets if you need them.

Sources of Information

Max Appeal
Telephone: 0800 389 1049 Website: www.maxappeal.org.uk
The Max Appeal is run by parents for parents, carers, their families and anyone affected by a 22q11 deletion syndrome. They run a family support group which holds regular meetings and runs age specific events throughout the year for families to get together. Their events are designed to make life enjoyable and our information aims to inform and help. They run a very comprehensive website which contains information and advice for parents and there is a series of downloadable "leaflets" covering a wide range of subjects.

Contact A Family
Helpline: 0808 808 3555 Website: www.cafamily.org.uk
The Contact a Family website is for families who have a disabled child and those who work with them or are interested to find out more about their needs. Contact a Family is the only UK charity providing support and advice to parents whatever the medical condition of their child. They have information on over 1,000 rare syndromes and rare disorders and can often put families in touch with each other.

Internet
The internet contains a wealth of information about medical conditions and treatments and users need to be confident that the information they read on the internet is reliable and accurate. The Judge Health website **www.judgehealth.org.uk** has guidelines that help you make informed decisions about Web sites and gives advice on how to search the internet for health information.

The development of this document has been a truly collaborative process. We would like to thank everyone who has contributed by sharing his or her experiences and knowledge especially:

North West Regional Genetics Service 22q11 Consensus Group

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