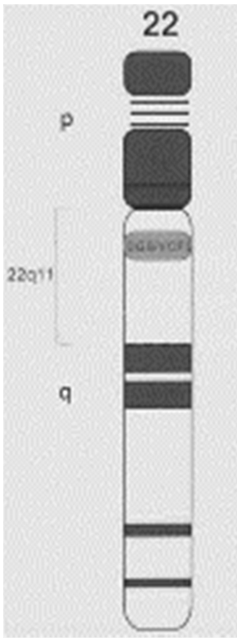
Information for Parents with a New-Born Baby

The purpose of this Information Leaflet is to inform parents about the issues that often show up at birth and early on in life. Your team should refer to you to other specialists as necessary.

What is DiGeorge/VCFS/22q11.2 deletion syndrome?



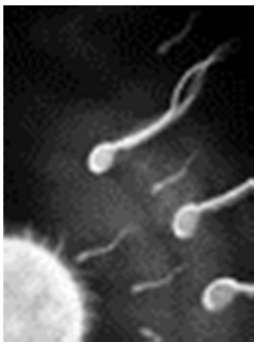
A syndrome is a collection of findings found occurring together in a pattern. Dr Angelo DiGeorge described one set of symptoms involving heart defects, endocrine and immunity issues. A speech pathologist called Dr Bob Shprintzen described another set of symptoms involving palate and heart problems, and subtle facial features, he called this Velo-Cardio-Facial syndrome.

The genetic name is 22q11.2 deletion (twenty two-q-one-one-point-two) syndrome, this means that there are some missing genes at a specific location on one of the 22nd chromosomes, it's like a genetic post-code. Shortened to "22q DS" for simplicity!

Why did this happen to us?

In about 85% of cases neither parent has 22qDS. There is a 'mistake' in the DNA copying process that results in some genes becoming lost, or deleted. This is a problem of the human race, a natural phenomenon, or just plain bad luck!

IT IS NO-ONE'S FAULT!



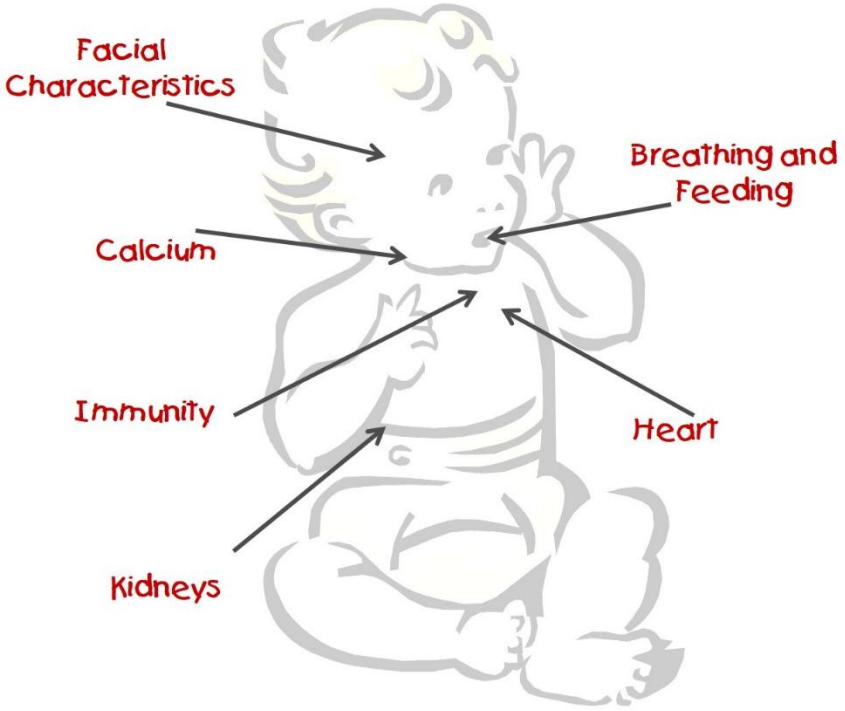
It is vital to understand that there is nothing that could have been done to cause this to happen or prevent it from happening. Your baby was destined to have this condition from the instant of conception. Either the egg or sperm from which the baby was made had those genes missing.

What does this mean for my baby?

Every baby with this condition is unique.

There really isn't a set pattern to it at all. The important thing to focus on is that with your love and support your baby can grow up to enjoy life as any other person.

The most frequently noted issues for babies are:



Facial Characteristics:

These are very subtle. Small, low set ears are noted, and a small jaw and mouth, sometimes without the bow shape to the top lip. A slightly broad bridge to the nose with small nostrils. Other things are a quiet cry and long fingers (hands of pianist!).

Feeding and Breathing:

Lots of babies with 22q have problems feeding, perhaps vomiting milk through the nose or just very slow to feed. This might be due to weak muscles or a palate problem. Sometimes fluids/feeds can 'go down the wrong way' this is called aspiration and can be dangerous.

Some babies have 'reflux' where feeds are regurgitated. It is very helpful to be seen by a feeding specialist or a speech and language therapist familiar with the condition usually through a cleft palate team. Tube feeding (through the nose or directly in to the stomach) is often recommended, and whilst this can feel alarming, it is often for a very short period of time and helps your baby through those potentially tricky early weeks and months.

Wheezing may be noticed from birth. This can be due to structural abnormalities in the throat or floppy air tubes in the throat or lungs. Referral to a respiratory specialist may be helpful.

Heart Defects:

These are usually of the main out-flow tracts of the heart, ie the aorta and pulmonary arteries, as well as the chambers of the heart (holes). If you don't have a cardiologist then a paediatrician can refer you for specialist tests.

Calcium:

This can cause 'tetany', a tremble in the arms perhaps, or seizures. Calcium levels can fluctuate but this is treated with medication, usually calcium and vitamin D supplements. There may be other endocrine (gland/hormone) issues.

Immunity:

Your baby may be more susceptible to chest and other infections. It is important that your baby is treated carefully and seen early on by an immunologist to establish how their immune system is working.

Kidney problems:

A missing kidney can be detected by ultra-sound scan. There may be other issues but these very rarely cause any problems but a review by a renal specialist may be sought.

Finally…

Our information is compiled from various resources including the Max Appeal Consensus Document on the Diagnosis and Management of 22q11.2 deletion. References and sources of evidence are freely available using the contact details above.

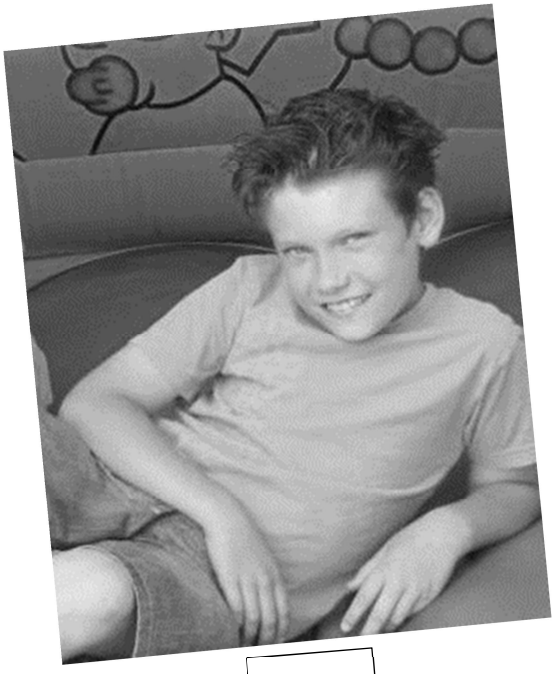
We are also grateful to our families, like Jessica's mum and dad for letting us use this picture of their lovely little girl.

Max Appeal has a range of information and advice leaflets covering these and many other aspects of 22qDS.

As your child grows other issues may well become apparent and Max Appeal is always there for you to call.

Feel free to get in touch at any time!

Some more of our 22 cuties:



Kieran

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