

Information for parents about the FISH test for 22q11 deletion syndrome

Before your child has pharyngeal flap surgery, he or she needs a blood test called the FISH test for 22q11 deletion syndrome.

Please take the requisition with you to the Out Patient Laboratory.

What is the FISH test?

FISH is the short term for the Fluorescent In Situ Hybridization test. This is a genetic test that will tell us if your child has the 22q11 deletion or not. The results give general information about your child's entire set of chromosomes as well as specific information about the long arm of chromosome 22 at a specific spot called "11".

With 22q11 deletion, a small piece of genetic information is missing. This is called a deletion. If your child's FISH test shows this deletion, the result is "positive" for 22q11 deletion syndrome.

What is 22q11 deletion?

22q11 deletion has many names. It is most commonly called Velocardiofacial syndrome (VCFS). VCFS is a genetic condition with a certain pattern of characteristics:

- **Velo** refers to the roof of the mouth (palate). The child may have speech problems or a cleft palate.
- **Cardio** refers to the heart. The child may have a heart defect.
- **Facial** refers to certain features of the face. The child may have slight differences in the eyes, ears or nose.

There are some other medical problems that may also be related to 22q11 deletion.

Why does my child need a FISH test?

A small percentage of children with hypernasal speech have 22q11 deletion syndrome. Of the children with the deletion, a few have displaced carotid arteries in their neck. These arteries work normally, but are in a different position than usual. They may be near the part of the throat where pharyngeal flap surgery is done.

Therefore, it is important to know before surgery if your child has this deletion.

If the FISH test shows that your child does have the deletion (a positive result), your child will need neck imaging to determine the exact location of the carotid arteries.

About 11% of patients tested at our clinic receive positive results. Occasionally another genetic abnormality is found unexpectedly. If either happens you will meet with a developmental pediatrician and a genetic counselor. At that time you can discuss what this might mean for your child and your family.