

What are the medical needs of children with the 22q11.2 deletion?

Once the diagnosis of a 22q11.2 deletion has been made, what should happen next? Since children with a 22q11.2 deletion may have problems in many different body systems, for example, heart, palate, and learning style, it makes sense that they should have a comprehensive evaluation of all areas which could be affected. At The Children's Hospital of Philadelphia, we recommend that all patients with a 22q11.2 deletion have the following evaluations, at least once, to rule out any potential problem. (This list is continuously changing as new findings are recognized amongst patients and is, therefore, not all inclusive. Also note that some problems, such as speech delay, for example, are not obvious at birth and can only be identified over time):

- * **Cardiology** - if the child does not have a known cardiac defect, an evaluation with a cardiologist is important, although the likelihood of finding a previously unrecognized problem is low. An evaluation may include a chest x-ray, electrocardiogram (study of the electrical impulses of the heart), and an echocardiogram (ultrasound of the heart).
- * **Child Development and Psychology** - children with a 22q11.2 deletion may have a difference in learning style requiring some extra help in school. In addition, young children may have delays in their developmental milestones and might benefit from early intervention, such as physical, occupational, and speech therapy. Therefore, it is important for patients with the deletion to have developmental testing on a regular basis to determine if there is a problem, and to identify appropriate intervention when needed.
- * **Cleft palate team** - all children with the 22q11.2 deletion should be seen by a plastic surgeon who specializes in cleft palate and VPI (velopharyngeal incompetence). In addition, they should be seen by a speech and language pathologist who can also assess speech, language, and feeding (in a young child). Some centers have specialized computer methods of objectively measuring speech to decide if intervention is necessary. Some teams also have growth specialists and dentists who monitor the growth of the face, jaw, and teeth.
- * **Endocrinology** - some children have problems with low calcium in the newborn period and require a calcium supplement. This is often handled by the cardiologist or pediatrician. If the problem continues, an endocrinologist (hormone doctor) may be asked to see the patient. In addition, some older patients with the 22q11.2 deletion have problems with growth (short stature) and would benefit from an evaluation by an endocrinologist since, in some cases, this is treatable.
- * **ENT and Audiology** - children with a palatal problem often have trouble with ear infections and can benefit from seeing an ear, nose, and throat doctor (ENT). Since some patients with a 22q11.2 deletion have hearing loss, a hearing test (audiogram) would often be beneficial. In addition, some children have problems with “noisy” breathing. These children in particular benefit from an evaluation by an ENT. This is often part of the cleft palate team evaluation.
- * **Feeding Specialist** - patients with a 22q11.2 deletion often have feeding difficulties, usually in the newborn period. These difficulties may be helped by having an evaluation with a pediatrician who specializes in feeding. This would only be recommended if a problem already existed.

- * **Genetics** - the geneticist is the most likely person to have an overview of the diagnosis. These would include keeping up to date with the new findings associated with the 22q11.2 deletion. Therefore, a yearly evaluation with genetics is often beneficial in answering questions regarding the patient's general care, recurrence risk, and the availability of prenatal diagnosis.
- * **Immunology** - many newborns with the 22q11.2 deletion have problems with their immune systems and, therefore, may have trouble with infections or trouble handling certain vaccines. Most children outgrow this problem by their first birthday, but some patients continue to have some trouble into later childhood and adulthood. We suggest that every patient be evaluated by the immunologist at least once.
- * **Neurologist** - rarely, children with a 22q11.2 deletion have symptoms such as a seizure disorder or problems with balance which require the services of a neurologist. We suggest that an initial evaluation with a neurologist may be helpful in some cases.
- * **Urology** - some patients with the 22q11.2 deletion have problems with their kidneys, including a missing kidney, which would be detected by a renal ultrasound. In addition, some children have difficulty with urinary tract infections, bedwetting or urinary frequency, and would benefit from seeing a urologist who is a specialist in this area.
- * **Other** - some patients with a 22q11.2 deletion commonly have: constipation, leg pain, and differences in the carotid arteries (blood vessels in the neck which lead to the brain). Other patients with the 22q11.2 deletion have problems which are seen fairly infrequently, these include: bony abnormalities such as extra fingers, extra ribs, a missing bone of the forearm, and problems of the vertebrae (spine) which sometimes leads to scoliosis (curvature of the spine); spina bifida, juvenile rheumatoid arthritis, eye problems, premature fusion of the "soft spots" or fontanelles (craniosynostosis), and emotional or psychological problems. Patients who have a problem in one of these areas are referred to the doctors who care for that particular problem.

22q and You

"Dedicated to Teaching, Learning, and Caring for patients with a 22q11.2 deletion"

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