ACPA Family Services
Resources for your cleft journey

INFORMATION ABOUT CROUZON SYNDROME (CRANIOFACIAL DYSOSTOSIS)

What is Crouzon Syndrome?

Crouzon syndrome, also called craniofacial dysostosis, is one of a large group of facial birth defects in which there is abnormal craniofacial fusion (joining between some of the bones of the skull and of the face). This fusion does not allow the bones to grow normally, affecting the shape of the head, appearance of the face and the relationship of the teeth. Crouzon was a doctor who described a patient with a characteristic group of deformities (syndrome) which were then observed in other individuals.

What causes it?

Crouzon syndrome is caused by an abnormality in the genes. If both parents are unaffected (showing no sign of the syndrome themselves), this abnormality is the result of a change in the genetic material at the time of conception. The exact cause of this change is not known. If one parent is affected, the abnormal gene is then known to have been contributed by that parent.

Does this mean that this can happen again in my family?

If both parents are unaffected, the chances of a second child being born with Crouzon syndrome are extremely small. However, if one parent is affected, the chances that any pregnancy will result in a child with this syndrome is 1 out of 2 (50% risk). For this reason, it is very important that both parents of an affected child be thoroughly examined before any recurrence risks are quoted to them.

If my child, who has Crouzon, marries and has children, will all the children have it too?

If your other children are unaffected (not showing signs of the syndrome), there is no increased risk to their children. If another family member has the syndrome, the occurrence risk for each pregnancy will be 50%.

Will my child be disabled?

There is no data to indicate that mental disability is a regular feature of this syndrome. Development should be evaluated periodically and if any concerns regarding mental function arise, appropriate referral for testing should be made.

What kinds of problems should I expect?

Like most birth defects, Crouzon syndrome varies in severity from patient to patient causing more problems in some than others. For a complete evaluation, optimum treatment planning, and comprehensive services, we advise you to contact a craniofacial center. At such a center, an experienced multidisciplinary staff composed of representatives from different healthcare specialties will assist you with care as well as in anticipating and meeting problems.

You should watch for any sign of ear disease and hearing loss, since research indicates that individuals with Crouzon syndrome may be quite vulnerable to ear problems. For an infant, the specialists at the craniofacial center can assess hearing in the early months of life.

Hearing should be monitored as your child grows.

While many children with Crouzon syndrome develop speech and language normally, some do not. The speech pathologist at the craniofacial center assesses speech and language development at regular intervals and will advise you if therapy is indicated.

Some individuals with Crouzon syndrome have problems with dry eyes, excessive tearing, or muscle balance (strabismus). Your child should receive a screening eye examination by an opthamologist (eye doctor) and any problems should be treated as soon as they arise.

The major problem for individuals with Crouzon syndrome is underdevelopment of the upper jaw. This produces facial deformity (bulging eyes and sunken middle third of face) and malocclusion (abnormal relationship between the upper and lower jaws). Dental and plastic surgery specialists monitor facial growth and correct deformities.

The facial deformity and need for treatment of Crouzon syndrome may create problems in family and social relationships, school placement, and so on. The craniofacial center may have a psychologist or social worker, or can refer you to one for evaluation and counseling if needed. Remember that children with Crouzon syndrome, like all other children, are individuals. They vary in social adjustment, academic achievement, and in their ability to cope with adults. The professionals at craniofacial centers try to maximize each child's potential by offering early diagnosis and treatment, when indicated.

Other specialties represented on the team vary somewhat from center to center. If your child has needs or problems requiring other specialties, you will be referred to them as needed.

What kind of treatment is available for my child?

The need, extent and timing for treatment of the deformities of Crouzon syndrome depend upon how severely the individual is affected and the age. For the infant, surgery may be required to release and reshape the bones of the skull, so that they may grow more normally. Orthodontics, to straighten the teeth, and jaw surgery, to place the teeth in a more normal position, may be done during childhood, teenage, or even adult years. There are complicated operations which are usually performed by specially trained craniofacial surgeons associated with major craniofacial centers.

What should I be doing for my child now?

First, be certain that the diagnosis is correct. Crouzon resembles several other syndromes, and not all physicians are aware of this. A geneticist can provide the necessary evaluation and information. Second, locate a craniofacial center. You may not have a center in your city but the care your child will receive will be more than worth the inconvenience of traveling to another city. Third, meet other individuals and families affected by similar facial differences by joining a parent-patient support group.

For more information:

This fact sheet and many others have been produced by:

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