

## **3 Diagnosis and referrals**

### **3.1 Antenatal diagnosis**

#### **3.1.1 Recommendations on making the diagnosis**

The fetal face can be studied with ultrasound very early in gestation. Examination of the fetal face should be a component of guidelines for second trimester ultrasound examination [6].

#### **3.1.2 Limitation of ultrasound**

Although the correct ultrasound examination technique should enable clefting of the upper lip to be identified, it is difficult to detect an isolated cleft palate antenatally [7]. What is often described as a 'cleft palate' on ultrasound may be just a cleft alveolus. Training of the professional undertaking the ultrasound diagnosis should include information on basic embryology of clefting and how treatment of clefts is managed.

#### **3.1.3 Suspected cleft lip and/or palate**

If a cleft has been suspected, there should be a referral to a specialist in ultrasound diagnostics to confirm the diagnosis. As a cleft lip and/or palate may be associated with other anomalies, early assessment and diagnosis is necessary.

Clear information about cleft lip and palate should be given to families if a cleft is suspected. Genetic counselling for patients and families should be available on request. The main topic for discussion is the association with other anomalies and the recurrence risk for following pregnancy.

If the cleft is part of a complex syndrome, specific information about it and about the fetus viability should be given to parents during the meeting.

In the absence of other anomalies, a fetus with a cleft does not require a change in standard obstetric care.

#### **3.1.4 Recommendations on referral to the cleft team**

The following recommendations should be followed:

- a referral from the antenatal diagnostics unit to a multidisciplinary cleft team and to a cleft support organisation (where it exists) should be made as early as possible after diagnosis. The referring unit should ensure it maintains up to date contact information on the local or regional cleft team(s);
- parents should be contacted by a member of the cleft team as soon as possible after receiving the referral; and
- parents should be offered a face to face meeting with a member of the cleft team within one month of the confirmed diagnosis.

The aim of the referral to the cleft team is to provide parents with support and counselling, giving clear information about treatment and successful management of cleft lip and palate. Families don't generally expect the diagnosis or know anything about cleft lip and palate and they have a lot of questions and concerns about bringing up a child with a cleft. Some families may be thinking about terminating the pregnancy.

Psychosocial support for parents who are expecting a child with a cleft should be available. Parents may need help in normalising the pregnancy and understanding that the cleft does not define the baby. The following topics may need addressing:

- grief adjustment process;
- possible feeling of guilt;

- impact on the family;
- how to handle reactions from others; and
- whether or not it will be possible to breast feed.

Parents should be given contact details for the cleft team and be informed that the team is able to provide additional information at any stage during the pregnancy. The information needs of families after a diagnosis of a cleft are provided in chapter 11.

## **3.2 Postnatal diagnosis**

### **3.2.1 Recommendations on making the diagnosis**

Diagnosis of a cleft palate is often missed. Proper assessment of the palate is of paramount importance:

- a palpation of the palate is not sufficient on its own. Routine assessment of all newborn babies should include a visual inspection of the palate with a torch and spatula to depress the tongue;
- every newborn baby with a cleft should be seen by an experienced paediatrician as soon as possible.

### **3.2.2 Recommendations on referrals**

Each maternity unit should designate a staff member to liaise with the cleft team to ensure that all staff are familiar with the contact arrangements and maintain an up to date knowledge and understanding of current practice in providing care for children with a cleft lip/palate.

Neonatal intensive care should be used only where specific needs justify it.

The maternity unit should provide basic information on cleft care such as general explanation of the condition, the reasons for treatment, treatment options, location of the cleft team, and details of support groups. This may be supplemented by leaflets, booklets or other kinds of information – websites, posters, and phone numbers.

Involvement of the cleft team around the birth will be necessary whether or not co-morbidities are present, such as Pierre Robin Sequence (see annex B).

All babies should be referred by relevant professionals to the cleft team as soon as possible after diagnosis. A cleft team member (a nurse specialising in cleft care where they exist) should visit at the earliest opportunity after receiving the referral. The team member will visit the mother at the maternity unit to observe the child during a feed, offer advice and supply any feeding equipment considered necessary.