

## 8

# Conclusions and recommendations

After thorough discussions of the many initial options, the following major themes were proposed:

## 8.1 Treatment of CFA

Three interrelated research issues were addressed within the clinical theme.

### 8.1.1 Evidence-based care

This issue focuses on the replacement of current widespread uncertainty and confusion in clinical care with a sound evidence-base derived from rigorous clinical research.

There is a pressing need to mobilize a critical mass of clinical research expertise and to access sufficiently large samples of patients for adequately-powered clinical trials. Initial efforts should include the following:

- trials of surgical methods for the repair of different orofacial cleft subtypes, not just unilateral clefts;
- trials of surgical methods for the correction of velopharyngeal insufficiency;
- trials of the use of prophylactic ventilation tubes (grommets) for middle-ear disease in patients with cleft palate;
- trials of adjunctive procedures in cleft care, especially those that place an increased burden on the patient, family, or medical services, such as presurgical orthopaedics, primary dentition, orthodontics and maxillary protraction;
- trials of methods for the management of perioperative pain, swelling and infection, and nursing;
- trials of methods to optimize feeding before and after surgery;

- trials addressing the special circumstances of care in the developing world in respect of surgical, anaesthetic and nursing care;
- trials of different modalities of speech therapy, orthodontic treatment and counselling.

Equally urgent is the need to create collaborative groups, or improve the networking of existing groups, in order to develop and standardize outcome measures. There is an especially urgent need for work on psychological and quality-of-life measures, and economic outcomes.

For rare interventions, prospective registries should be established to hasten collaborative monitoring and critical appraisal, equivalent to Phase I trials. Relevant topics would be craniostylosis surgery, ear reconstruction, distraction osteogenesis for hemifacial macrosomia and other skeletal variations, midface surgery in craniofacial dysostosis, and correction of hypertelorism.

### **8.1.2 Quality improvement**

Quality improvement focuses on the development and dissemination of methodologies for monitoring and improving the delivery of clinical services.

The international adoption of a set guideline for the provision of clinical services and for the maintenance and analysis of minimum clinical records of cleft care is proposed. Various registries of clinical outcomes have recently emerged and are working independently. Efforts should be made to harmonize these.

### **8.1.3 Access and availability**

Identify strategies to maximize access to adequate levels of care for all affected individuals, irrespective of nationality.

In large parts of the world, routine public health services are unable to afford treatment for CFA. Three general approaches can be identified: high volume indigenous centres of excellence; contracts between non-governmental organizations (NGOs) and local hospitals; and volunteer short-term surgical missions. The long-term benefit of these efforts could be developed by:

- a survey of the charitable organizations involved and the scale of their work;
- an appraisal of the cost-effectiveness and clinical effectiveness of the different models of aid;

- the promotion of dialogue between different NGOs to develop commonly-agreed codes of practice and adoption of the most appropriate forms of aid for local circumstances, with an emphasis on support that favours indigenous long-term solutions;
- the initiation of clinical trials concerning the specifics of surgery in a developing country setting, one-stage operations, optimal late primary surgery, anaesthesia protocols (e.g. local anaesthetic, inhalation sedation), antisepsis;
- the development of common core protocols for genetic, epidemiological and nutritional studies alongside surgery.

## **8.2 Gene/environment interaction**

### **8.2.1 Epidemiology**

The overall conclusions to be drawn from the data presented are as follows:

- there is ample evidence of the distinctly different nature of CL/P and CP, and emerging evidence of distinct differences in sub-groups within these overall conditions;
- there is a great deal of geographical variation which is more apparent for CL/P than CP;
- there is considerable variation in the proportion of cases of OFC with additional congenital anomalies and syndromes;
- it is evident that migrant groups retain rates of CL/P similar to those of their area of origin;
- there is no consistent evidence of time trends, nor is there consistent variation by socioeconomic status or seasonality, but neither of these aspects have been adequately studied;
- there is considerable international variation in the frequency of orofacial clefts, but validity and comparability of data are adversely affected by numerous factors, among which are:
  - (a) source population of births considered (hospital versus population),
  - (b) time period,
  - (c) method of ascertainment,
  - (d) inclusion/exclusion criteria, and
  - (e) sampling fluctuation;
- there are many parts of the world where we have little or no information on the frequency of OFC, in particular parts of Africa, Central Asia, Eastern Europe, Middle East and Russia.

### **8.2.2 Etiology**

The following points are relevant:

- there are multiple genes involved in OFC;
- analysis should be separated for CL, CL/P and CP as CL/P is not the same as CL only;
- heterogeneity should be expected and therefore different populations will need to be examined for validation of a result;
- nutrition remains an eligible area for research, and the roles of folic acid and multivitamins, including folic acid, vitamins A, B2, B6 and B12, as well as zinc, need further investigation;
- smoking, alcohol, epilepsy, certain medications and environmental factors may explain a small but appreciable portion of birth defects;
- main gaps in knowledge are examination of co-teratogens and gene/environment interaction e.g. with alcohol are there co-teratogens, such as folate deficiency, and is there a threshold beneath which alcohol is safe?

It is important to be able to differentiate the exposure and the genetic predisposition; and identify those at risk to allow selective counselling since general advice regarding alcohol and smoking in relation to disease is not easy to impart in attempting to achieve changes in behaviour.

One major issue in the reporting of associations with exposures is the distinct possibility of publication bias in the literature.

### **8.2.3 WHO aims and objectives for gene/environment interaction research**

The ultimate humanitarian and scientific research objective in CFA birth defects is *primary prevention*.

The WHO project aims to:

- provide support for planning and development of research protocols that will advance understanding of etiology and inform future prevention initiatives;
- facilitate internet-based research databases;
- support gene/environment interaction studies with international standardization of research protocols to inform the design of future efforts towards primary prevention.

These objectives can be achieved by:

- the reinforcement of existing research collaborations, and
- the setting up of new research collaborations.

#### **8.2.4 Future research challenges**

With the availability of the human genome sequence, researchers have increasing opportunities to study the role of genes and GEI in human health and disease. Such opportunities come with major challenges, in three main areas:

- **The first area relates to data:** to identify and, if possible, rank the major data gaps separating our current knowledge from that needed for clinical and public health action.
- **The second area relates to methods:** how to conduct, analyse and present studies of multiple genetic and environmental factors in ways that efficiently fill the data gaps.
- **The third area relates to people and institutions:** how to learn more and more quickly using the unique opportunities inherent in international collaboration.

Common core protocols for data collection and further studies into research methodology to compare various data analysis models are urgently required.

### **8.3 Genetics**

The focus of the genetics component of the WHO Craniofacial Conference was on discussing those technologies, analytic approaches and populations that will best move us forward towards a better understanding of the etiologies of craniofacial abnormalities with particular reference to those that have strong genetic components. While recognizing that the environment and stochastic events play an important and, often, major role in predisposing to craniofacial anomalies, in many situations the role of genetics is compelling.

#### **8.3.1 Phenotype/genotype correlation**

- A number of specific single-gene disorders with recognizable Mendelian inheritance, including some holoprosencephaly and craniosynostosis syndromes, serve as benchmarks for ways in which gene identification can proceed from clinical description and family-based studies through traditional cloning and functional analysis.

- The definition of non-syndromic cleft lip and palate remains ambiguous, and new gene discoveries leading to improvements in genetic diagnoses will potentially improve sensitivity and specificity of genotype/phenotype correlation.
- There is some emerging evidence that traditional separations between cleft lip, with or without cleft palate, and cleft palate only, may be breaking down, and further work in this area is essential.
- It is therefore important in research to be able to sub-phenotype cases of children whose abnormalities are limited to clefts, or clefts and one additional abnormality. Clinical descriptors that will allow breaking this group down into finer detail will be particularly important in facilitating genetic analysis.

### **8.3.2 Analytical methodologies**

- Technological and analytic approaches will include new methodologies for genotyping, the strategy by which markers will be chosen for genotyping, and the selection of candidate genes when that approach is being utilized.
- The strengths and weaknesses of traditional linkage approaches versus affected pedigree-member approaches and transmission disequilibrium testing (TDT) and linkage disequilibrium were also developed.
- The strengths of these approaches often overlap and combinatorial approaches using candidate genes in conjunction with affected pedigree-member linkage and TDT can all be carried out in parallel with one another.

### **8.3.3 Collection and storage of genetic data**

- Analysis is driven by sample collection, and there are both strengths and weaknesses in:
  - (a) rapid, cost-efficient, and small-amount sample collection, as is exemplified by blood spots or cheek swabs; and
  - (b) whole blood or cell line collections that would allow for more extensive analysis of protein and RNA.
- International collaboration is essential in that etiologies are likely to be diverse across populations but with some underlying gene and environmental causes shared in common.
- Multi-centre collaborations afford the opportunity for the collection of large numbers of samples to have sufficient power to confirm

linkage or association studies; there are a number of active on-going collaborations.

### **8.3.4 *Parallel research and multidisciplinary approach***

- The role of animal models and the insights gained from developmental biology into choosing both genes and pathways involved in CFA genetics have never been more apparent than they are now.
- It will be through the interactive efforts of clinicians, epidemiologists, statisticians, molecular biologists and developmental biologists that we will make our most rapid progress.

### **8.3.5 *Role of the World Health Organization***

In the ongoing efforts to globalize CFA research, the WHO group will coordinate work on outlining candidate genes, markers, analytic approaches and animal models of use, and will streamline efforts towards establishing collaborative groups to establish a set of protocols and guidelines for future efforts in this arena.

## **8.4 Prevention**

### **8.4.1 *Primary prevention***

Orofacial clefts appear to have substantial environmental causes; the potential for their occurrence thus seems considerable. The pattern of occurrence of orofacial clefts is different from that of neural tube defects so their causes may also be different.

- **Maternal tobacco** use has been consistently associated with a modest elevation in risk of orofacial clefts but the attributable risk may be of public health importance. Moreover tobacco use is rapidly increasing among women, especially in technologically developing countries, and many women are exposed to passive smoking in the home and workplace.
- **Maternal alcohol** use, well known as a cause of the fetal alcohol syndrome, has also been associated with risk of isolated orofacial clefts in some, but not all, studies. The type and context of alcohol consumption differs considerably across populations and more consistent methods are needed for the assessment of maternal alcohol intake. The possible increased risk of orofacial clefts and other CFA related to the common exposures of smoking and alcohol use during pregnancy is a message that should be incorporated into health promotion programmes for women of reproductive age.

- **Maternal nutritional factors** have been associated with the risk for orofacial clefts in human population studies, although strong evidence of a causal relationship is still lacking. The most promising candidate nutrients include folic acid and pyridoxine (vitamin B-6) and some evidence also exists of possible roles for riboflavin (vitamin B-2) and vitamin A.

#### **8.4.2 *Intervention trials***

The current state of equipoise regarding maternal nutrition and orofacial clefts makes intervention trials of specific nutrients an urgent priority. The proven intervention of folic acid supplements in the prevention of occurrence of NTDs must also be acknowledged in the design of prevention trials involving folic acid. No single trial is likely to be definitive and trials are needed in diverse populations in both industrialized and technologically developing countries. Trials in high-risk populations are more likely to detect a treatment effect than trials in low-risk populations, and at lower cost and with greater speed.

#### **8.4.3 *Choice of nutrient***

The choice of specific nutrient interventions should be based on prior detailed studies of biochemical indicators of nutritional status in the population of interest, and all prevention trials should adhere to current ethical and methodologic standards. Poorly conceived and conducted trials are unethical because they waste limited resources and add further delay to discovering effective interventions.

#### **8.4.4 *Recurrence trial***

An orofacial-cleft recurrence-prevention trial is far more feasible than a trial of prevention of primary occurrence, but would still require many thousands of high-risk mothers. Orofacial cleft surveillance systems and registries in countries around the world need to be further developed and linked to provide the critical infrastructure for orofacial-cleft prevention trials.

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# Annex 1:

## European Collaboration on Craniofacial Anomalies (EUROCRAN)

### Background

In 2000 a partnership of 14 European centres was awarded funding under the European Commission's Framework V Programme for research to carry out the EUROCRAN project. EUROCRAN, which will run for four years – between 2000 and 2004 – brings together researchers from a range of clinical/scientific disciplines with the shared aim of improving the management and understanding of craniofacial anomalies (CFA). This will be achieved through five inter-related work packages (*see Annex 2*).

### Participation

The work described in the work packages will be achieved through the development of common core protocols and with the involvement of participating centres from the European Union, the European Economic Area and the states of Central and Eastern Europe.

If you would like to participate or require more information please contact:

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### **Further materials compiled by EUROCRAN is included as follows:**

- Annex 2: Work packages
- Annex 3: Policy statements
- Annex 4: Practice guidelines
- Annex 5: General principles governing record-taking (provisional)

# Annex 2:

## Work packages

### **Work package 1: Surgical trial**

A multi-centre randomized trial of the primary surgery for infants with complete unilateral cleft lip and palate will compare four surgical methods in three concurrent trials. Infants will be randomized to a surgical method common to all three trials or the usual local method. Surgeons will do an approximately equal number of their usual method and the common method according to the randomization scheme maintained at the trial coordinating centre.

### **Work package 2: Gene/environment study**

A population-based multi-centre case-parent triad study to investigate gene/environment, and gene/gene interactions and genetic susceptibility polymorphisms operating in the etiology of orofacial clefting (OFC) will be carried out. Mothers with affected babies who are participating in the study will complete a structured interview regarding diet and other exposures in the periconceptual period. In addition samples will be taken from the mother, father and child for DNA extraction and genotyping. Gene variant analysis will then be carried out to investigate the interaction between:

- (a) maternal nutritional factors and maternal/fetal metabolism genes;
- (b) genes coding for xenobiotic metabolism enzymes and environmental teratogens;
- (c) developmental genes (growth factor genes, homeobox genes) and environmental factors.

### **Work package 3:**

## **A chromosomal approach to identifying OFC genes**

A cohort of European patients with OFC associated with apparently balanced chromosomal rearrangements will be identified and their breakpoints/clinical phenotypes catalogued. A bank of immortalized cell lines will be established from a sub-set of these patients where two or more instances of a specific breakpoint has been associated with OFC. Both high throughput molecular cytogenetic techniques and available sequence data from the Human Genome Project will be used to identify genes that have been interrupted by two or more breakpoints. These genes will be fully characterized and screened for mutations and polymorphisms that may be used in Work Package 2.

### **Work package 4:**

## **Molecular diagnosis of monogenic craniofacial anomalies**

The aim is to develop sensitive molecular assays for the mutations underlying a number of craniofacial malformation syndromes using Treacher Collins Syndrome (TCS) as a paradigm. This expertise will be disseminated to other molecular laboratories in the EUROCRAN group such that it will be available on a local basis.

### **Work package 5:**

## **Directory of resources**

A European Craniofacial Anomalies Directory of resources for European teams will be created. The Directory will include:

- a register of clinical teams, their reported clinical protocols and research interests, governmental and non-governmental agencies involved in the treatment and research of CFA, European CFA surgical missions to developing countries, model research protocols and examples of successful grant applications;
- a dynamic database/website of emerging data from Work Packages 2 and 3 such as chromosomal breakpoints, candidate genes and study protocols;
- a "good practice" set of clinical records for consecutive cases of OFC including cephalometric radiographs, dental casts, photographs and speech samples so that teams can compare local outcomes to the reference set;
- a prospective registry of complex treatment outcomes using distraction osteogenesis as an exemplar.

## Annex 3:

# Policy statements

- (1) The professional involved in cleft care should provide basic information on cleft care and on the proposed treatment to any potential patient and/or patient's guardian. Basic information should contain at least:
  - a general explanation of the condition, the reasons for treatment, what may or may not be achieved, the stages of treatment including examination, record collection and general protocols – this may be supplemented by leaflets, booklets or other kinds of information;
  - an explanation of why a specific treatment is considered necessary for the individual patient, what specifically is involved: method, timing, duration cost, what the specific goal is and possible side effects.
- (2) When a treatment is considered, the professional engaged in cleft care should take into consideration the desires and attitudes of the patient and/or those of the patient's guardian. The professional should also pay attention to and inform the patient/patient's guardian of the risks and benefits inherent in the potential alternative treatment options, including no treatment or no further treatment.
- (3) If requested, it is the professional's responsibility to provide a procedure for obtaining a second opinion for the patient. If requested, this procedure should be communicated to the patient before treatment starts.
- (4) After an episode of treatment, the professional engaged in cleft care should inform the patient and/or patient's guardian on:
  - outcome of treatment relative to the defined goal;
  - undesirable effects of treatment;
  - expected future development.
- (5) The professional engaged in cleft care should analyse and document any complaints or praise expressed by the patient and/or the patient's guardian.
- (6) The professional engaged in cleft care should give consideration to the burden of the treatment. Considerations should include financial as well as non-financial burden, such as treatment duration, effort from the patient and/or patient's guardian and discomfort as a result of treatment.

- (7) During the process of treatment, the professional involved in cleft care should continuously evaluate treatment progress against the planned treatment and act accordingly.
- (8) Organizations and institutes responsible for the provision of cleft care should:
- encourage the cleft professional to follow the policies described above and to acknowledge the patient's rights;
  - recognize and encourage the professional's right to provide treatment that can be expected to improve the patient's condition whilst minimizing adverse effects;
  - recognize and encourage that decisions on treatment priority should be based on criteria proposed by the cleft professionals in consultation with the patient and/or patient's guardian. This is especially so in a situation with insufficient treatment resources;
  - recognize and encourage that access to treatment should not depend on the patient's ability to pay;
  - recognize that cooperation of the patient with the advice and instructions of the cleft professional is necessary in order to achieve a successful result.

# Annex 4:

## Practice guidelines

### Part I: Health-care needs

- (1) **Neonatal emotional support and professional advice:** In the event of prenatal diagnosis and as soon as possible after the birth of a child with a cleft, parents should be given emotional support and advice about the child's future management by a specialist in cleft care.
- (2) **Neonatal nursing:** Difficulties in feeding are common in the early days of life and specialist advice on feeding should be provided.
- (3) **Surgery:** Primary surgery to close clefts of the lip and/or palate should be performed by an experienced and qualified surgeon according to a protocol agreed by the team. Further corrective procedures may be necessary for some patients in later years and should be performed by an experienced and qualified surgeon according to a protocol agreed by the team.
- (4) **Orthodontic/orthopaedic treatment:** For children with cleft lip and palate orthodontic/orthopaedic treatment should be available when necessary and should be performed by an experienced orthodontist.
- (5) **Speech and language therapy:** Early assessment of speech and language problems, advice to parents and the availability of corrective therapy by an experienced speech and language therapist should be provided.
- (6) **Ear, nose and throat (ENT):** ENT problems should be identified at an early stage and the necessary therapy should be provided.
- (7) **Clinical genetics/paediatric developmental medicine:** As cleft lip and/or palate may be associated with other anomalies early assessment and diagnosis is necessary. Genetic counselling for patients and families should be available.
- (8) **Emotional support and professional advice for the growing child and its parents:** Emotional support and professional advice for parents, patients and their environment is often necessary and should be available.
- (9) **Dental care:** Regular dental care should be available.
- (10) **National register:** A national register should be in place for accurate recording of children born with cleft lip and/or palate and related craniofacial anomalies.

## Part II: Organization of services

- (1) Cleft care should be provided by a multidisciplinary team of specialists.
- (2) Members of the team should have special training in cleft care.
- (3) The team should agree on the stages of treatment including the examination, record collection and general protocols.
- (4) There should be one person responsible for quality improvement and communication within the team.
- (5) Coordination of the care of individual patients is important since numerous specialities are involved. This should be the responsibility of one member of the team.
- (6) The number of patients referred to the team should be sufficient to sustain the experience and specialist skills of all team members and to allow evaluation/audit of the team's performance within a reasonable period of time. It has been recommended that cleft surgeons, orthodontists and speech therapists should treat at least 40-50 new cases annually. However, it is recognized that individual member states have the right to provide care for their own population.

## Part III: Finances

Resources should be available to cover the following care for children with cleft lip and palate:

- (1) Emotional support and professional advice during the neonatal period.
- (2) Neonatal nursing.
- (3) Surgery.
- (4) Orthodontic/orthopaedic treatment.
- (5) Speech and language assessment and therapy.
- (6) Ear, nose and throat treatment.
- (7) Clinical genetics/paediatric developmental medicine.
- (8) Emotional support for the growing child and its parents.
- (9) Travel expenses.
- (10) General dental care including cleft related prosthodontics.

# Annex 5:

## General principles governing record-taking (provisional)

### 1. Records for treatment planning/monitoring

- Clinical records should be taken for individual patients to allow treatment planning, monitoring treatment progress and treatment evaluation.
- The timing and nature of these records will depend on the clinical protocols followed by individual teams.
- Treatment and associated record-taking protocols should be agreed and clearly set out by the cleft team.

### 2. Records for quality improvement/research

Additional records may be taken for a number of other reasons:

- follow-up of a series of patients to provide an overview of the outcome of care;
- to allow retrospective comparisons of different protocols;
- as part of a prospective clinical trial with ethical approval;
- as part of an agreed protocol for intercentre quality-improvement comparisons or comparison against known standards;
- as part of an agreed research protocol;
- other reasons, such as medico-legal, second opinion.

### 3. Safeguards

- Exposure of patients to unnecessary radiation should be avoided.
- Research and quality-improvement records should only be taken when there is an established written protocol on how they will be put to use.
- Research and quality improvement records should not be taken without the consent of the patient/parent/guardian.
- Research and quality improvement records should coincide as far as possible with the records for treatment planning/monitoring (statement 1 above).

## 4. Timing of minimum records

**Table 1: Complete cleft lip and palate (UCLP & BCLP)**

Timing	Models	Lateral skull radiograph	Photographs	Speech/ tympanometry	Audiometry	Patient/parent satisfaction
Primary surgery	✓		✓			
3 years				✓*	✓*	
5/6 years	✓		✓	✓	✓	
10 years	✓	✓	✓	✓	✓	
18+ years	✓	✓	✓	✓		✓

\* = If hard palate is closed.

**Table 2: Cleft palate only**

Timing	Models	Lateral skull radiograph	Photographs	Speech/ tympanometry	Audiometry	Patient/parent satisfaction
Primary surgery	✓		✓			
3 years				✓	✓	
5/6 years	✓			✓	✓	
15/16 years	✓	✓	✓	✓	✓	✓

**Table 3: Cleft lip only**

Timing	Models	Photographs	Patient/ parent satisfaction
Primary surgery	✓*	✓	
3 years			
5/6 years	✓*	✓	
10 years			
18+ years		✓	✓

\* = Only in cases with cleft of the alveolus as well as cleft lip.

**Table 4: Alveolar bone grafting**

Timing	Intra-oral x-ray	Photographs
Just before bone graft	✓	✓
6 months after graft	✓	
After canine fully erupted	✓	✓

**Table 5: Pharyngoplasty**

Timing	Speech sample
Just before operation	✓
One year after operation	✓

**Table 6: Orthognathic surgery**

Timing	Lateral cephalogram	Models
Just before operation	✓	✓
One year after operation	✓	✓

## 5. Record-taking methodology (provisional)

Discussion of the precise method of record taking is continuing. The following however, provide a suggestion that is currently being used widely in Europe.

### 5.1 Photographs

**Background:** The vast majority of surgeons and orthodontists use still photographs for documentation of clefts. Very few clinicians use video recording of clefts pre- or post-operatively. If photographs of clefts which appear in any publication are examined it is clear that there is no uniformity or standardization of the way in which such photographs are taken. For comparative studies the following views are recommended.

#### Basic views to be taken:

- Frontal, both laterals, inferior (columellar) view.
- Three-quarter ( $\frac{3}{4}$ ) facial (oblique) view.

#### Dynamic views:

- During smiling and whistling – in the cooperative older patient, these views will give an idea of function of the circum-oral musculature.
- Video recording will be better for assessing circum-oral movement but this will also need to be standardized and cannot be used routinely at present.

#### Lighting and background:

- Lighting for the studio should be two fill-in lights and the main light synchronized with the camera. In the ward or operating theatre a single flash unit is appropriate.
- The background should be blue.

### Framing of the picture:

- For frontal view, the camera should be set at a ratio of 1:8.
- For lateral view, the camera should be set at a ratio of 1:8.
- For inferior view, the camera should be set at a ratio of 1:4.

### Camera and lens:

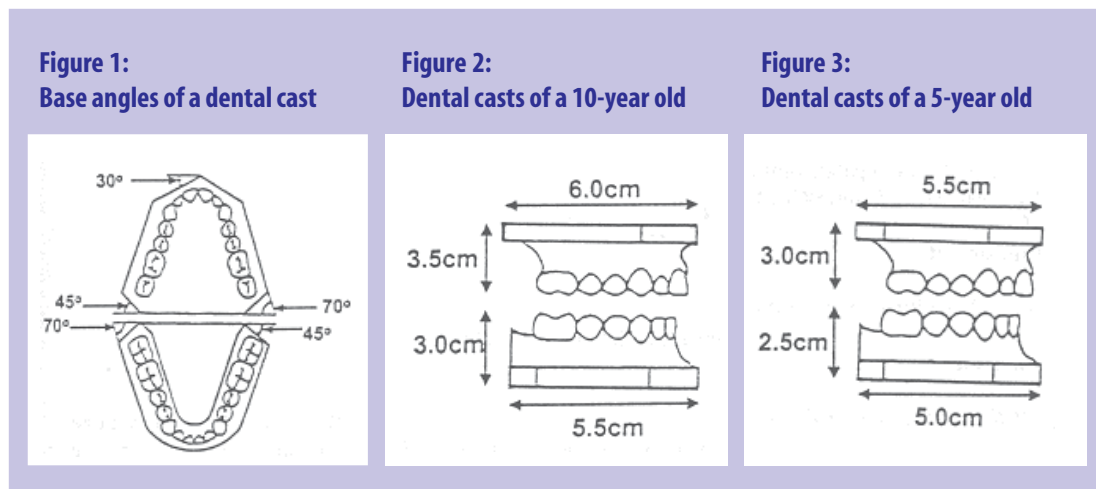
- Suggested camera is Nikon F3 with a 105mm lens or equivalent.
- Film type and speed need not be standardized.

## 5.2 Dental casts

**Background:** Dental casts need to be made from well-taken impressions which include all teeth, the palate and the buccal sulcus. For comparative studies the casts need to be prepared in a standard manner so that the source of the models cannot be identified.

**Preparation:** Models should be:

- cast in vacuum-mixed white stone, for example Crystacal R;
- hand trimmed, using a fine wheel to the standard heights and angles shown in Figures 1-3 below;
- finished with wet and dry paper (not soaped).



## 5.3 Speech

**Background:** A fundamental problem for speech and language pathology has been the lack of an acceptable framework for measuring speech. Various groups have proposed procedures for measuring, recording and reporting speech data cross-linguistically, but to date there is no one recognized method.

Proposals have come from Henningsson and Hutters (1997), and also from Dalston, Marsh, Vig, Witzel and Bumstead (1988). In Britain, Sell, Harding and Grunwell (1994) developed the Great Ormond Street speech assessment (GOS.SPASS) tool. This is now a nationally-agreed speech

assessment tool for cleft palate and/or velopharyngeal incompetence in English. From GOS.SP.ASS, Razzell, Harding and Harland (1987) devised the Cleft Audit Protocol for Speech (CAPS), a more succinct protocol specifically designed for audit purposes.

**Ages:** 3-4 years; 5-6 years; 10 years; 15-16 years (cleft palate only); 18+ years (UCLP and BCLP)

**Equipment:** A good quality audio recording using a high quality microphone.

**Variables:**

- **Intelligibility:** a rating should be made upon spontaneous speech. The CAPS scale can be used to judge how "understandable" a persons speech would be to familiar and unfamiliar listeners (there are however flaws with this method).
- **Nasality:** the presence/absence and degree of hypernasality, hyponasality, audible nasal emission and nasal turbulence can be judged and rated on a five-point scale (see CAPS). An agreed instrumental method for assessing nasality has yet to be recommended.
- **Assessing articulation:** set sentences and single words containing consonant sounds in different word positions (beginning, middle and end) should be repeated, for example "Bob is a baby boy" or equivalent in the native language, and recorded for CAPS. Targeted sounds are\*: p, b, f, n, t, d, s, ʃ, tʃ, dʒ, k, g.

Errors made can be broadly categorized or grouped according to CAPS:

- front of mouth oral-sound errors;
- back of mouth oral-sound errors;
- non-oral sounds;
- passive errors;
- immaturities.

**References:**

- Henningsson G, Hutter B. Perceptual assessment of cleft palate speech with special reference to minimum standards for intercentre comparisons of speech outcome. In: Lee ST, Huang M, eds. *Transactions 8th International Congress on Cleft Palate and Related Craniofacial Anomalies*. Stamford Press Pte Ltd: Singapore 1997.
- Dalston M, Marsh JL, Vig KW, Witzel MA, Bumstead RM. Minimal standards for reporting the results of surgery on patients with cleft lip, cleft palate, or both: A Proposal. *Cleft Palate Journal*, 1988; 25: 3-7.
- Sell D, Harding A, Grunwell P A. Screening assessment of cleft palate speech (Great Ormond Street speech assessment: GOS.SP.ASS). *European Journal of Disorders of Communication*, 1994; 29: 1-15.
- Harding A, Harland K, Razzell R. *Cleft Audit Protocol for Speech (CAPS)*. Available from K Harland, Speech and Language Therapy Department, St. Andrew's Plastic Surgery Centre, Stock Road, Billericay, Essex CM12 OBH, United Kingdom, 1987.

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\* Depending on the speech sound in each language, but should contain plosives, fricatives and a nasal consonant (p, b, t, d, k, g, f, s, n).