Subjective and objective aspects on living with a cleft. A study focused on the isolated cleft palate and the total bilateral cleft lip and palate

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To my grandmothers
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Abstract

Aims: To gain a deeper understanding of the social life aspects in individuals treated for isolated cleft palate (iCP) or total bilateral cleft lip and palate (BCLP). Furthermore, to study risks of having other birth defects in relation to the length of the iCP. For the BCLP, dental conditions and treatment results were evaluated. Finally, opinions on the treatment results for BCLP were compared between orthodontists and young adults.

Subjects and Methods: First a qualitative interview using Grounded Theory was carried out. Twelve persons with iCP or BCLP participated. The length of the iCP was evaluated on dental cast models and the presence of other birth defects were studied from records for 343 persons. Dental conditions with focus on the anterior maxillary dentition were studied in 35 persons with BCLP. A web-based questionnaire, with intra and extra-oral photos from 12 of these persons, was evaluated by a group of 25 orthodontists and 20 young adults without any type of cleft.

Results and conclusions: In the qualitative interview study, seven important categories were revealed. The main category was “hoping to be like others”. This feeling was not only related to extra-oral appearance. One or more other birth defects can be seen in almost every third child with an iCP. Congenital heart disease and intellectual disability were the most common. A total iCP implied a significantly higher risk (1.7 times) for other birth defects compared to a partial iCP. There was a high frequency (40 %) of missing and/or peg-shaped lateral incisors. This was a factor that affected the treatment results in that a symmetrical upper frontal dentition was hard to achieve during orthodontic treatment. A good sagittal occlusal relationship and a positive overjet were achieved for a majority of the young adults with BCLP. The esthetic outcome of treatment results was more negatively rated by the group of young adults compared to the orthodontists. Additionally, individual opinions on each set of photographs were independent, regardless of the category of evaluators, in explaining the rating scores.

Key words: isolated cleft palate, bilateral cleft lip and palate, social life, additional malformations, dental treatment outcome, esthetic assessment

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Original papers

This thesis is based on the following papers, which will be referred to in the text by their Roman numerals:


**Abbreviations**

<table>
<thead>
<tr>
<th>Abbreviation</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>AIC</td>
<td>Akaike Information Criterion</td>
</tr>
<tr>
<td>BCLP</td>
<td>Bilateral cleft lip and palate</td>
</tr>
<tr>
<td>CI</td>
<td>Confidence interval</td>
</tr>
<tr>
<td>CIA</td>
<td>Confidence interval analysis</td>
</tr>
<tr>
<td>CLP</td>
<td>Cleft lip and palate</td>
</tr>
<tr>
<td>Cont.</td>
<td>Continue</td>
</tr>
<tr>
<td>ENT</td>
<td>Ear, nose and throat</td>
</tr>
<tr>
<td>iCP</td>
<td>Isolated cleft palate</td>
</tr>
<tr>
<td>IRF6</td>
<td>Interferon regulatory factor-6</td>
</tr>
<tr>
<td>MTHFR</td>
<td>Methylenetetrahydrofolate reductase</td>
</tr>
<tr>
<td>N/A</td>
<td>Not applicable</td>
</tr>
<tr>
<td>PiCP</td>
<td>Partial isolated cleft palate</td>
</tr>
<tr>
<td>PiCP(h)</td>
<td>Partial isolated cleft palate involving the hard and soft palate</td>
</tr>
<tr>
<td>PiCP(s)</td>
<td>Partial isolated cleft palate involving only in the soft palate</td>
</tr>
<tr>
<td>PR</td>
<td>Pierre Robin Sequence</td>
</tr>
<tr>
<td>PVRL1</td>
<td>Poliovirus receptor like-1</td>
</tr>
<tr>
<td>QOL</td>
<td>Quality of life</td>
</tr>
<tr>
<td>RR</td>
<td>Relative risk</td>
</tr>
<tr>
<td>TBX22</td>
<td>T-box transcription factor-22</td>
</tr>
<tr>
<td>TGFA</td>
<td>Transforming growth factor alpha</td>
</tr>
<tr>
<td>TGFβ3</td>
<td>Transforming growth factor beta 3</td>
</tr>
<tr>
<td>TiCP</td>
<td>Total isolated cleft palate</td>
</tr>
<tr>
<td>TPR</td>
<td>Total population register</td>
</tr>
</tbody>
</table>
Subjective and objective aspects on living with a cleft.  
A study focused on the isolated cleft palate and the total bilateral cleft lip and palate.

Introduction

Background

Cleft lip and palate (CLP) is the most common craniofacial anomaly. CLP is categorized into different groups. The classification system varies depending on the purpose (Shprintzen, 2002). Extra-oral clefts (one-sided or bilateral) involve the upper lip and often the base of the nose. The extra-oral cleft can be combined with a cleft in the alveolar process with or without involving the palate. The severity of the condition and difficulty in treatment of the same type of cleft varies. The inherited growth pattern is a factor that may affect the final treatment outcome. The bilateral cleft lip and palate (BCLP) is a cleft that involves the base of the nose, the upper lip, and the alveolar process through the hard and soft palate (Fig. 1A, 1B). The isolated cleft palate (iCP) is not affecting the facial appearance since it only involves a secondary palate. It can either be a total or a partial cleft. The total iCP is a palatal cleft with an origin directly behind the foramen incisivum region. It extends all the way back through the hard and soft palate. A partial cleft palate sometimes partly involves the hard palate, and extends all through the soft palate. At times, only the soft palate is affected by the cleft.

![Figure 1A](image1a.png)  ![Figure 1B](image1b.png)

**Figure 1A.** An extra-oral photograph of BCLP.  
**Figure 1B.** Dental cast model of BCLP.

CLP related research has been pushing forward for decades. However, there is still little known about etiology, the life situations for persons born with a cleft and so on.
**Etiology**

BCLP or iCP could be either a feature in syndromes (Table 1) or isolated ones, so-called non-syndromic cleft. Causes of non-syndromic CLP still remain unclear (P. Mossey, et al., 2009). Many studies have suggested a relation between some maternal habits during the first tri-semester of pregnancy and the development of a cleft. There is strong evidence that maternal smoking is linked with an increased risk of having CLP (Wyszynski, et al., 1997; Little, et al., 2004). Regarding nutrition, multivitamin supplements are not confirmed as being a protective factor (Loffredo, et al., 2001; Goh, et al., 2006). Some animal studies have shown that a nutrient deficiency of folic acid and Zinc increased the risk of having a cleft (Warkany & Petering, 1972; Bienengraber, et al., 2001; Malek, et al., 2004). Exposures to some chemical substances; for example, agricultural chemicals, retinoids (Vitamin A derivatives), corticosteroids, some anticonvulsants drugs e.g. diazepam, carbamazipine, phenobarbital, or a virus infection also increases the risk for clefts and other congenital malformations (Dolovich, et al., 1998; Park-Wyllie, et al., 2000; Acs, et al., 2005; Romitti, et al., 2007).

**Table 1.** List of the congenital malformations commonly or occasionally featured with either CLP or iCP (Shprintzen, 2002).

<table>
<thead>
<tr>
<th>Syndrome</th>
<th>CLP</th>
<th>iCP</th>
</tr>
</thead>
<tbody>
<tr>
<td>Craniofrontonasal syndrome</td>
<td>common</td>
<td>common</td>
</tr>
<tr>
<td>Del (18p)</td>
<td>common</td>
<td>common</td>
</tr>
<tr>
<td>Ectrodactyly-Ectodermal dysplasia-Cleft lip and palate syndrome</td>
<td>common</td>
<td>common</td>
</tr>
<tr>
<td>Filliform adhesions with clefting syndrome</td>
<td>common</td>
<td>common</td>
</tr>
<tr>
<td>Niikawa-Kuroki syndrome</td>
<td>common</td>
<td>common</td>
</tr>
<tr>
<td>Oculoauriculovertebral spectrum</td>
<td>common</td>
<td>common</td>
</tr>
<tr>
<td>Opitz syndrome</td>
<td>common</td>
<td>common</td>
</tr>
<tr>
<td>Popliteal pterygium syndrome</td>
<td>common</td>
<td>common</td>
</tr>
<tr>
<td>Trisomy 13</td>
<td>common</td>
<td>common</td>
</tr>
<tr>
<td>van der Woude syndrome</td>
<td>common</td>
<td>common</td>
</tr>
<tr>
<td>Cri du chat syndrome</td>
<td>occasional</td>
<td>common</td>
</tr>
<tr>
<td>Escobar syndrome</td>
<td>occasional</td>
<td>common</td>
</tr>
<tr>
<td>Fetal Alcohol syndrome</td>
<td>occasional</td>
<td>common</td>
</tr>
<tr>
<td>Hay-Wells syndrome</td>
<td>occasional</td>
<td>common</td>
</tr>
<tr>
<td>Syndrome</td>
<td>CLP</td>
<td>iCP</td>
</tr>
<tr>
<td>----------------------------------</td>
<td>-----------</td>
<td>-----------</td>
</tr>
<tr>
<td>Rapp-Hodgkin syndrome</td>
<td>occasional</td>
<td>common</td>
</tr>
<tr>
<td>Treacher Collins syndrome</td>
<td>occasional</td>
<td>common</td>
</tr>
<tr>
<td>Wolf-Hirschhorn syndrome</td>
<td>occasional</td>
<td>common</td>
</tr>
<tr>
<td>Cryptophthalmos syndrome</td>
<td>occasional</td>
<td>occasional</td>
</tr>
<tr>
<td>Down syndrome</td>
<td>occasional</td>
<td>occasional</td>
</tr>
<tr>
<td>Oculodentodigital syndrome</td>
<td>occasional</td>
<td>occasional</td>
</tr>
<tr>
<td>Robinow syndrome</td>
<td>occasional</td>
<td>occasional</td>
</tr>
</tbody>
</table>

Genetic factors are also important. In non-syndromic CLP, some growth factors and metabolic enzymes such as TGFA, TGFß3 and MTHFR have been intensively studied (Wong & Hagg, 2004; Zeiger, et al., 2005; Vieira, 2006; P. Mossey, et al., 2009; Jagomagi, et al., 2010). Hereditary patterns have been described for families with syndromes e.g. the van der Woude syndrome. The gene IRF6, TBX22 and PVRL1, which causally associates with syndromes, also links to an occurrence of a non-syndromic CLP (Carinci, et al., 2007; Park, et al., 2007; Ferrero, et al., 2010). It leads to a hypothesis that any gene which relates to any syndromic CLP could possibly induce the risks of having non-syndromic CLP (Wong & Hagg, 2004). Spontaneous mutations are likely to keep the incidence values for rare syndromes, with clefts as a feature, at a rather stable level throughout decades. The research in gene-disease association is still a wide challenging field for future research.

**Epidemiology of CLP**

From the available data, cleft lip with or without cleft palate was reported at high rates in some parts of Latin America, China, and Japan. It was found in low rates in Israel and Southern Europe. Regarding iCP, the rates were high in Canada, some Scandinavian countries, but low in Latin America. South Africa had low rates of both types of cleft (P. Mossey & Castilla, 2001; P. Mossey, et al., 2009). However, the number of iCP might be underestimated since the disease is not externally visible. The global data is hardly comparable due to several reasons. The differences in collecting methods, sample sources, inclusion/exclusion criteria, and others, restrict the comparability (Gundlach & Maus, 2006; P. Mossey, 2007).

**Cleft related problems: Feeding, ear infections and speech development**

A person born with a CLP encounters problems from birth. Breathing problems are common in a combination of an iCP and an underdeveloped mandible, the Pierre
Robin sequence (Howard, 2002). A connection between oral and nasal cavity reduces the sucking ability. Feeding problems or general weakness may disturb normal growth and development. Instructions and exercises of feeding position and using a special feeding bottle might be needed (Sidoti & Shprintzen, 1995; Reid, et al., 2007). When the feeding problems are severe, a special feeder such as a naso-gastric tube and syringe may be advised. Recurrent ear infections are common and associated with pain, loss of hearing and delayed speech development (Sheahan, et al., 2002; Sheahan, et al., 2003; Flynn, et al., 2009).

The cleft involving the palate mostly affects speech to various degrees. The Velopharyngeal function and the length of the soft palate are important factors that relate to speech problems (Witzel, 1995). Often there are problems in constructing a soft palate long enough to prevent the air flow passing from the pharyngeal area up to the nose, thereby affecting the speech. A small partial palatal cleft is easier to treat than a very extensive cleft. Sometimes the speech is affected, but most often young adults develop a normal speech. However, speech training and additional surgical corrections may be needed in order to normalize the speech (Bardach, 1995).

Despite being complete or partial, the BCLP is considered to be the most challenging type of cleft, in order to receive a good result from both an esthetical point of view and in terms of developing a normal speech.

Social and psychological impact

The family situation is obviously different from that of a family with a healthy infant. When a newborn infant is diagnosed as having a cleft, several different emotions may possibly come into existence. Shock, anxiety, guilt and/or disappointment are common feelings (Beaumont, 2006). Inappropriate family adaptation could psychologically affect children with CLP. Parents sometimes turn to protective behaviors (Collett & Speltz, 2007).

Living in a city or rural area can make a difference on how other people address a person who has been treated for a CLP. Parents, family, friends, teachers and the school system are factors in a society that may have influence on a person’s self-esteem. Social interactions are other factors related to a person’s appearance (Turner, et al., 1998; Endriga & Kapp-Simon, 1999). Compared to children without craniofacial conditions, children with CLP received less positive responses and avoided longer conversations with their friends (Kapp-Simon & McGuire, 1997). They showed problems, reported by parents or teachers, with depression, anxiety, unhappiness and behavioral problems (Millard & Richman, 2001; Hunt, et al., 2007). A Norwegian study, by Ramstad et al., found that depression and anxiety were frequently reported by persons with CLP. Fewer people with CLP were married and the marriages occurred later in life (Ramstad, et al., 1995a). A later review article, published in 2005, described that children and adults with clefts, in general, did not experience severe
psychological problems, but for extra-oral clefts, they expressed low satisfaction with their facial appearance (Hunt, et al., 2005).

There is individual variation in adjusting to living with a visible deviation. Some manage to cope but others become psychologically distressed (Moss, 1997; Stavropoulos, et al., 2010). A similar situation of living with a visible deviation appears with persons with CLP. They could, to some extent, have difficulties regarding the severity of psychological problems. Some studies have suggested that persons with treated CLP showed no evidence of severe psychological problems (Hunt, et al., 2005). However, it is obvious that even in the completely treated cases of CLP, the scars remain visible. In Gothenburg, psychological consultations are available whenever needed in order to prevent any negative impacts. The iCP is the type of cleft most frequently associated with other birth defects (Stoll, et al., 2000). Congenital birth defects are also likely to bring a negative experience into life.

**Dental factors**

The outcome of dental and orthodontic treatment is important in that the esthetic look of the dentition is an important part of a person’s facial appearance. Maxillary dental midline deviations and asymmetric dental arch forms were found in children with CLP (Semb & Shaw, 2001). Anterior crossbite has been reported as common (Friede & Katsaros, 1998).

It is known that clefts of the primary palate may interfere with normal tooth development, and hereditary patterns of having or missing peg-shaped laterals are shown among family (Ranta, 1986). The morphology of the canine is usually normal, but regarding the position, it is frequently impacted. Canine impaction, often in a palatal position, occurred 10 times more in persons with complete CLP than in non-clefts (Semb & Schwartz, 1997). A variation in number, morphology, enamel formation and eruption of the teeth in the cleft area can make it a challenge to create good esthetics together with good functions (Boehn, 1963; Dahl & Hanusardottir, 1979). Well aligned teeth with a correct midline help in harmonizing the face with the symmetry of the upper frontal teeth being especially important.

A high frequency of posterior crossbite in younger children was reported (Ross, 1987; Reisberg, 2000). The reasons for this could be developmental maxillary deficiencies in persons with CLP, functional imbalance due to the presence of the cleft, and postoperative scar tissue formation (Ross, 1987). In a recent study by Mohlin et al., it was found that in persons who did not have a CLP, the negative functional consequences from crossbite were rare (Mohlin, et al., 2007).

**The Gothenburg CLP team and the Gothenburg treatment protocol**

In Sweden, all newborns with CLP are registered. They are regularly followed with a standardized registration and treatment protocol for each type of cleft until 19
years of age. No private care is available and the Gothenburg CLP team takes care of all children with clefts in the south-western part of Sweden. The team consists of plastic surgeons, orthodontists, speech pathologists, a psychologist and specialized nurses. Other medical professionals such as audiologists, ENT-specialists and pediatricians are consulted when needed. At times, local speech pathologists and orthodontists are asked to help with the treatment, in collaboration with the team, when the patient lives too far away for frequent visits. The treatments and registrations with records, dental cast models, X-rays and photographs follow standard protocols according to the type of cleft.

The surgical treatment for all persons with a BCLP, in this study, started with a lip adhesion at three months of age followed by soft palate closure at four to six months (Lilja, 2003; Friede & Figueroa, 2007). However, if the cleft was very wide, a lip adhesion was performed on each side at different times. At least 3 months later, the operation was carried out on the other side of the cleft lip and combined with the closure of the soft palate. A bilateral lip-nose repair was performed at twelve months of age. Delayed hard palate closure protocol was performed and the procedure has gradually been done at an earlier age (Friede, et al., 1980; Lilja, 2003). The simultaneous bone grafting is considered when a child enters the period of mixed dentition (Lilja, 2003). Bone grafting is performed on one side at a time.

The treatment protocol for all persons with an iCP was somewhat different from persons with a BCLP. The soft palate was first closed around six months of age or at least during the first year of life. Later, when the residual cleft of the hard palate had spontaneously narrowed, the hard palate closure was performed. The procedure has gradually been performed at an earlier age (Lilja, 2003).

The idea of the research plan was to start exploring for general information on the experiences the persons had in their life situations and their remembered feelings when they grew up. The information from the first study was hoped to generate ideas and hypotheses for studying other factors that could enlighten other aspects on living with a CLP.
Aims

The general aim of this thesis was to study the subjective and objective aspects of living with CLP. Two different types of clefts were chosen; iCP and BCLP. The iCP is not visible from an extra-oral view. The treatment of BCLP is very challenging in order to achieve good facial and dental appearance. This implies that the variables studied were different for the two types of clefts. A comparison between clefts was not intended. The specific aims for the four studies were:

- To explore a deeper understanding about social life aspects in individuals treated for iCP or BCLP.
- To study risks of having other birth defects in relation to cleft morphology in terms of the cleft length of individuals with iCP.
- To evaluate dental conditions and treatment results, primarily focused on the anterior maxillary dentition, in treated BCLP.
- To compare opinions of orthodontists with those of young adults without any type of cleft, concerning treatment results in persons with BCLP.
Subjects and Methods

Subjects

The study population in all four studies consisted of Caucasian persons born with either an iCP or a total BCLP. They had been registered for treatment at the Sahlgrenska University Hospital, Gothenburg.

Study I: An interview study with a qualitative approach on social aspects of young adults with CLP

Persons older than 18 years of age with either an iCP or a total BCLP participated in the study. The participants were randomly chosen from a total sample of the region’s registered persons with CLP. By the end of the study, when the data reached saturation, there were 12 participants (6 males, 6 females). The ages ranged between 24–33 years of age. Five of them had a BCLP and seven had an iCP.

They all resided in the area of the Västra Götaland Region and had fulltime employment.

Study II: A descriptive study of cleft extension and other birth defects in iCP

Data was collected from all Caucasian children who were born in the Västra Götaland Region with an iCP. There were, in total, 343 children (171 males, 172 females) born with an iCP during 1975–2005. Submucous clefts were not included.

Study III: A descriptive follow-up study of dental conditions with focus on the anterior maxillary dentition in young adults with BCLP

The study group was a total sample of children registered with a total BCLP. They were born between the years 1975 to 1991 and living in the Västra Götaland Region. Inclusion criteria were to be of Caucasian origin and to have followed the treatment and registration protocol with available dental cast models. In total, there were 35 persons who fulfilled the criteria and had cast models taken at 13, 16 and 19 years of age. Persons with a Simmonart’s band were included. A Simmonart’s band; defined as a string of tissue with a length of 5 mm or less, diagnosed as having a total cleft. This band connects the central and lateral parts of the cleft lip directly below the nose on one or both sides.
Study IV: A web-based questionnaire study on esthetic views of facial and dental appearance in BCLP. A comparison between the professional and non-professional evaluators

Persons with BCLP were randomly selected from the participants in Study III for a web-based questionnaire. They had to have a complete set of photographs after finishing their orthodontic treatment at ages 16 to 19 years. Twenty-four sets of photographs were available. Out of these, one person with a very good treatment result and one with a poor result were chosen. The reason was to have diversity among the photo cases. Ten cases were then randomly chosen out of the remaining 22. Finally, the web-based questionnaire consisted of 12 cases.

The evaluators of the photo cases were a group of 25 randomly chosen specialists in orthodontics. They were working in the Västra Götaland Region. The other group consisted of 20 young adults older than 18 years of age. They did not have any type of cleft. All the young adults had experiences from their own orthodontic treatment with fixed appliances. They were asked to participate when visiting the Department of Orthodontics. The mean age was 19.2 years (range 18–23 years). Among the orthodontists, the age varied from young recent graduates up to experienced orthodontists, close to retirement.

<table>
<thead>
<tr>
<th>All newborns with any type of CLP</th>
<th>iCP and BCLP</th>
<th>Caucasian criteria</th>
<th>iCP</th>
<th>BCLP</th>
</tr>
</thead>
<tbody>
<tr>
<td>Study I</td>
<td>Study II</td>
<td>Study III</td>
<td>Study IV</td>
<td></td>
</tr>
<tr>
<td>N = 12</td>
<td>343</td>
<td>35</td>
<td>12</td>
<td></td>
</tr>
</tbody>
</table>

**Figure 2.** Diagram of subject recruitment and number of participants (N) in each study.
Methods

Study I: An interview study with a qualitative approach on social aspects of young adults with CLP

The qualitative interview technique used was based on the Grounded Theory (Strauss & Corbin, 1998). Persons with an iCP or a BCLP were initially contacted by letter. They were asked if they were willing to participate in a telephone interview. This meant that they would have enough time to talk about their social life for 45 minutes in an in-depth interview. The interview was open-ended and tape recorded. All participants were asked about their lives from when they were young up till their present age. After finishing an interview, the record was transcribed verbatim into text by the interviewer for analysis. A new participant was added first after pre-analyzing each interview until the information reached saturation. All data was analyzed following the Grounded Theory technique using open and selective coding processes. The core category and other categories were identified to generate the hypothesis of the study.

Study II: A descriptive study of cleft extension and other birth defects in iCP

Data from dental cast models and patient records was collected. The cast model used was the first one taken before the first surgical session or pre-surgical treatment with a plate. Surgery was performed when the infants were around 6 months old and the cleft in the soft palate was corrected. Each cast model was checked to see if the diagnose in the patient’s record was matched. The length of the iCP was measured in order to classify whether it was total (TiCP) or partial (PiCP). The partial clefts were sub-grouped into two classes; PiCP(h) when the cleft partly involved the hard palate, and PiCP(s) when only the soft palate was involved. Dental cast models of both total and partial iCP are shown in Fig. 3.

![Figure 3. Dental cast models of TiCP [A], PiCP(h) [B] and PiCP(s) [C].](image-url)
Sources of information were the patient records from the plastic surgery clinic, orthodontic clinic and speech therapy clinic of the Sahlgrenska University Hospital. The information collected is listed in Table 2. The number of total live births in the region was acquired from the Total Population Register (TPR) in order to calculate the incidence values of iCP. The incidence values and the estimates of relative risks were calculated by CIA program (Morris & Gardner, 2000).

**Study III: A descriptive follow-up study of dental conditions with focus on the anterior maxillary dentition in young adults with BCLP**

Patient records, dental cast models and photographs were analyzed following a standardized protocol for the study. The main interests were dental treatment outcomes focusing on the upper anterior teeth, including orthodontic and prosthetic treatment. Maxillofacial surgery was also recorded. The detail information is shown in Table 2.

The surgical treatment protocol for this period of time was 1) lip adhesion at 3 months, 2) soft palate closure at 6 months, 3) first lip and nose repair at 12-18 months, 4) hard palate closure and/or bone grafting (one side at a time) at 8-9 years of age.

The statistics with percentage, median values and ranges were calculated for the different variables.

**Study IV: A web-based questionnaire study on esthetic views of facial and dental appearance in BCLP**

The web-based questionnaire consisted of an introduction and a sample set of photos, 12 pages of cases with BCLP, and a page to select whether the evaluator was an orthodontist or a young adult. A complete set of photographs consisted of extra-oral photos of the front and profile, and intra-oral photos of the right, left, and frontal view. All photos were cut off beneath the eyes to make them anonymous.

The first question concerned the first three things the evaluator noticed when looking at the photos. For the second question, the following topics were rated; 1) the profile of the face, 2) the form of the upper lip, 3) the regularity of the upper teeth, 4) the regularity of the lower teeth, 5) the shape of the upper teeth, 6) the color of the upper teeth, and 7) the overall impression. There were four written alternatives to choose among; bad, fairly good, good and excellent. Finally, own comments could be added.

The answers were compared between the professional and non-professional evaluators. The first things noticed by both the orthodontist and young adult groups are presented in percentage. The logistic regression with Akaike Information Criterion values (AIC), and Pearson’s chi square test were calculated to find if there were any significant differences either between the categories of evaluators or associations between these two groups.
A summary of topics of interest, types of research and techniques for all four studies is presented (Table 2). All studies were approved by the Regional Ethical Committee of the University of Gothenburg.

**Table 2. Summary of the four studies; type of study, topics of interest and techniques used.**

<table>
<thead>
<tr>
<th>Study</th>
<th>Type of study</th>
<th>Topics of interest</th>
<th>Technique</th>
</tr>
</thead>
<tbody>
<tr>
<td>I</td>
<td>Qualitative interview analysis: Grounded Theory</td>
<td>Childhood experiences, school life, treatment experiences, meaning of the diagnosis, social participation, future planning</td>
<td>Open ended, telephone interviews of persons older than 18 years of age</td>
</tr>
<tr>
<td>II</td>
<td>Descriptive research</td>
<td>Cleft diagnosis, length of cleft, gender, other associated malformations, PR, ear infections</td>
<td>Data collection from patient records and dental cast models before initial surgery of the palate</td>
</tr>
<tr>
<td>III</td>
<td>Descriptive research, a follow-up study</td>
<td>Appearance of the maxillary anterior teeth (overbite, overjet, tooth form, missing teeth, facial and dental midline, intra-maxillary symmetry, prosthetic treatment, maxillofacial surgery).</td>
<td>Data collection from patient records, dental cast models and photographs focused on 13, 16, 19 years of age</td>
</tr>
<tr>
<td>IV</td>
<td>Questionnaire</td>
<td>Facial appearance, dental appearance, other individual comments</td>
<td>Web-based questionnaire answered by orthodontists and young adults older than 18 years of age</td>
</tr>
</tbody>
</table>
Results

Social life experiences (Study I)

The interviews were carried out until the data reached saturation. Finally, twelve participants, seven with an iCP and five with a BCLP, participated in Study I. Difficult lives with clefts were revealed. The results showed seven important categories (Fig. 4) with “Hoping to be like other people” considered as the core category.

![Diagram](image)

**Figure 4.** The relation between the core category and other categories defined from the interviews for young adults with iCP or BCLP.

All seven categories were identified from many inconvenient situations expressed by the persons with treated iCP or BCLP. They described their feelings of being different from other people. Speech difficulty and deviated facial appearance were two major obstacles of living a normal life. Persons with iCP were affected by the
speech problem even after their speech had improved from training and/or surgery. Their memories from childhood still worried them. Persons with BCLP were both affected by present or previous speech problems together with their facial appearance such as scars, a deviating form of the upper lip and nose. Some of their difficult circumstances were bullying at school, difficult treatment visits, public speaking, job interviews, dating and starting a relationship. Persons who could not cope with their feeling of inferiority would end up with low self-esteem. However, “receiving recognition from significant others” was a category that reflected on how this problem could, after all, be solved. Persons who were married or had partners felt they had received recognition. They felt as if they were just as valuable as others and focused less on their condition. Their self-esteem increased. Those who had children described how they worried during their pregnancies that their child also would have a CLP.

**Cleft morphology (Study II)**

There were 343 Caucasian children who were born with iCP during 1975–2005 in the Västra Götaland Region (Table 3). The incidence value was 0.64/1000 live births.

Table 3. Sample characteristics from Study II.

<table>
<thead>
<tr>
<th>Sample characteristics</th>
<th>Number</th>
</tr>
</thead>
<tbody>
<tr>
<td>Total children born between 1975 and 2005</td>
<td>535,320</td>
</tr>
<tr>
<td><strong>Male</strong></td>
<td>275,737</td>
</tr>
<tr>
<td><strong>Female</strong></td>
<td>259,583</td>
</tr>
<tr>
<td>Total children born with iCP</td>
<td>343</td>
</tr>
<tr>
<td><strong>Male</strong></td>
<td>171</td>
</tr>
<tr>
<td><strong>Female</strong></td>
<td>172</td>
</tr>
<tr>
<td>Diagnosis</td>
<td></td>
</tr>
<tr>
<td><strong>TiCP</strong></td>
<td>34</td>
</tr>
<tr>
<td><strong>PiCP(h)</strong></td>
<td>161</td>
</tr>
<tr>
<td><strong>PiCP(s)</strong></td>
<td>137</td>
</tr>
<tr>
<td><strong>Missing</strong></td>
<td>11</td>
</tr>
</tbody>
</table>

Thirty-four percent of all subjects had other birth defects. From the statistical analysis, a significant difference between the length of the cleft and the risk of having other birth defects was found. Persons with a TiCP had a 1.7 times higher risk of having an additional malformation compared to those who had PiCP (RR=1.71,
CI=1.20-2.46). Among these birth defects, congenital heart disease and intellectual disabilities were the most common (19% and 18%). Nineteen different syndromes were revealed with the majority of Down’s syndrome and 22q11 deletion syndrome. 15% of the children born with iCP had the Pierre Robin sequence (PR). 43% had reports of ear infection episodes (Fig. 5).

![Figure 5. Percentage of children born with iCP associated with other health problems. * Not applicable information.](image)

**Dental treatment outcome (Study III)**

The data from dental cast models, patient records and photographs of 35 persons with BCLP showed that unilateral or bilateral missing incisors were common (40%) as were peg-shaped laterals (40%). Presence of an upper canine in the area of a missing lateral was more common than having a lateral implant. All except one person with a missing lateral incisor were treated so that the upper canine was placed in the same position on both sides. This means that in cases with only one missing lateral incisor, the contra lateral incisor was extracted. Only half of the persons with BCLP achieved an excellent occlusal relationship in all three dimensions; sagittal, vertical and transversal. The number increased to 57% when considering only normal sagittal/vertical relationships. 60% of the subjects showed good symmetry and a straight midline between jaws.

The most frequent treatments that persons with BCLP received were an early correction of the upper anterior teeth, an early treatment with transverse expansion, and active early treatment with fixed appliances (13 years of age). The values were 82, 58 and 55% respectively (Fig. 6).
Early correction of the upper anterior teeth
Early treatment with transverse expansion
Active early treatment with fixed appliances*
Orthodontic treatment of canines**
Retention phase***

Figure 6. Frequencies (%) of orthodontic treatments in 35 person with BCLP.
* Active early treatment with fixed appliances in persons at 13 years of age.
** Orthodontic treatment of repositioning canines in the area of missing or extracted lateral incisors on both sides.
*** Number of patients at 16 years of age in retention phase after orthodontic treatment.

External assessment on esthetics (Study IV)

The group of orthodontists and the group of young adults focused on different features in the facial and dental appearance of the persons with BCLP (Fig. 7).

For both groups, the upper lip was chosen among the three first noticed items. Almost half of the orthodontists first noticed the upper lip when they saw the set of photos, while the young adults gave their attention to the teeth. According to question two in the questionnaire, it was found that concerning the overall appearance, both groups of orthodontists and young adults assessed in a similar way. However, when it came to the consideration of dental appearance, the orthodontists were more positive than the other group in every issue including teeth alignment, shape/form and color of the upper teeth. The logistic regression analysis showed that according to the ACI value, individual assessments on different sets of photographs were independent of the group category of the evaluator. The results revealed the same pattern in every issue of question two.
Figure 7. Percentage of first noticed items reported by orthodontists and young adults.
Discussion

The cleft study project started with the qualitative interview in order to learn about the subjective feelings of living with a cleft (Study I). The study group consisted of individuals who had repairs of either total BCLP or iCP (total or partial). It was aimed for a large variety of clefts because, in this type of research, diversity theoretically increases the explanatory power of a phenomenon (Strauss & Corbin, 1998). Grounded Theory used in Study I is hypothesis generating. It is believed to be valuable in providing new information on different factors that are related to “living with a cleft”. A study consequently emerged on how others perceived the appearance of persons having undergone complete treatment for BCLP (Study IV). Research methods used were Grounded Theory approach, descriptive study and questionnaire study.

The study population represents the population with CLP born in the western part of Sweden. Two very different types of CLP were selected. BCLP (Study I, III, IV) represents the cleft with most extensive problems and the highest severity of facial deviation and dental problems. iCP (Study I, II) represents the cleft with no problems regarding facial appearance. However, speech impairment may exist.

The qualitative study revealed a core category “hoping to be like other people”. The other categories were “being treated differently from others”, “experiencing deviations from others”, “regarding oneself as being different from others”, “lack of recognition”, “low self-esteem” and “receiving recognition from significant others”. Marcusson et al. compared adults treated for CLP and persons without clefts of a similar age. Well-being and social life were poorer in persons with CLP although the overall QOL was high (Marcusson, et al., 2001). A study done in adolescents also showed that persons with CLP tended to be an outsider of daily conversations or at the periphery of a group (Kapp-Simon & McGuire, 1997). In a Norwegian study, anxiety and depression were more frequently reported. Persons with CLP married later in life and fewer were married (Ramstad, et al., 1995a). The feeling of appearing different from others was described both for the time period of growing up and as a young adult. It was an issue regardless of the type of cleft. The iCP is not visible since it is only located in the palate. However, speech problems before and after surgery are not uncommon as are recurrent ear infections. Priester et al. found that in 30-50% of Dutch toddlers with surgically treated CLP, the hyper nasality and conductive hearing loss remained as problems (Priester & Goorhuis-Brouwer, 2008). In order to improve the speech, several sessions with surgery may be needed along with speech training.
The total bilateral cleft is very challenging when the aim is to finally receive good facial and dental appearance. During growth, the BCLP is likely to affect the appearance of the nose since the columella often is short. There are bilateral scars that are more or less visible on the lip. The position of the premaxilla can be deviating. Many sessions of surgery, speech training and orthodontic treatment are needed in order to receive final good function and esthetics. The facial appearance in BCLP may be more deviant in a younger child than when older. The speech may also be more affected in a younger child who has not completed the treatment of the cleft. In Study I, bullying was frequently reported during the interviews. The children need a lot of support from family, teachers and psychologists. Baker et al. revealed an importance of having support from family and friends in that it reduced a negative family impact (Baker, et al., 2009). It may also be helpful to send out professional expertise to a school and create an understanding by teaching about clefts.

The Crouzon syndrome implies cranial disfigurement with a more or less deviating facial appearance. In a recently published qualitative interview study, young adults with the syndrome were asked how they handled their life situation. It was found that many of them reported coping strategies such as having engaging activities and avoiding exposed situations (Stavropoulos, et al., 2010). A questionnaire study done by Cochrane & Slade (1999) revealed the patterns of coping strategy associated with emotional adjustment (Cochrane & Slade, 1999). The adjustment process of persons with a deviation connected to the way they interpret their appearance and their self (Thompson & Kent, 2001). In Study I, “receiving recognition from significant others” was the coping strategy that increased a person’s self-esteem.

The iCP is the type of cleft most often associated with other birth defects (Milerad, et al., 1997; Hagberg, et al., 1998; Stoll, et al., 2000). One idea was to further study the risks for other birth defects since increased knowledge in this matter is important. The number of visits to the hospital or other institutions could have psychologically influenced the children. Another problem is the heavy burden for the whole family. Problems with other birth defects were not reported by any of the participants in the qualitative interview (Study I), but many persons described that they had to stay away from school due to hospital visits, speech training, dental treatment, consultations and other appointments. This made them feel “different from others”. From clinical experience, a hypothesis in Study II was found that a TiCP is more often correlated to other birth defects than a PiCP. The results showed that the risk was 1.7 times higher for a TiCP compared to a PiCP. No other published studies have been found concerning cleft length and risks for other birth defects.
Thirty-four percent of children with iCP had at least one other birth defect. The most common birth defects were congenital heart disease and mental disorders (Study II). In other studies, frequencies of malformations ranged from 22% to 71% (Shprintzen, et al., 1985; Milerad, et al., 1997; Shaw, et al., 2004; Vallino-Napoli, et al., 2006; Stoll, et al., 2007). A congenital heart disease was also reported as the most common isolated associated malformation in the study by Milerad et al. (Milerad, et al., 1997). Newborns with BCLP have also been reported to have an increased risk of additional malformations (Hagberg, et al., 1998). Repeated episodes of ear problems related to infections were noted in the records for almost 50% of the children. It is in line with another study where 45% of the children with cleft lip and/or cleft palate had a history of middle ear disease (Sheahan, et al., 2003). It is very important to inform the parents that ear infections are more common in clefts that involve the palate. One reason is malfunction of the Eustachian tubes. Recurrent ear infections may turn into chronic infections. Hearing is then decreased for longer periods with the development of speech being delayed in a young child.

The outcome of dental and orthodontic treatment is important in that the esthetic look of the dentition is an important part of the facial appearance. In the qualitative interview study, one category was “receiving recognition from significant others”. A nice smile is important and well aligned teeth help in harmonizing the face. The interaction between persons and the way they address others are among other factors relating to how a person looks (Trulsson, 2003). Study III was aimed at an objective evaluation and quality control of how the upper anterior dentition appeared after treatment of the young adults with BCLP, since a good symmetry of the upper frontal dentition and a correct midline between jaws helps in harmonizing the face (Semb & Shaw, 2001). Therefore, focus should be set on the appearance of the upper front, the symmetry, the size of the teeth on right and left sides, the alignment, the color of the teeth and the overall periodontal condition and dental caries.

The region for the upper lateral is often associated to esthetic problems in that peg-shaped laterals or missing laterals are common (each 40%). Very few implants were recorded but the development of smaller implants and good surgical techniques for bone augmentation makes it an option for the future. Replacement with the upper canines in the position of a missing or extracted lateral incisor may be a good treatment alternative. In order to create symmetry, the lateral incisor on the other side could also be extracted. However, it is important to be aware of the fact that eruption problems for the upper cuspids are common. Semb and Schwartz reported that canine impaction occurred 10 times more often in children with complete CLP compared to non-clefts (Semb & Schwartz, 1997). A palatal position for the canine was common and one reason suggested was scar tissue formation in the palate (Semb & Schwartz, 1997).
Study IV was aimed at investigating how other people perceived the facial and dental looks of young adults treated for a BCLP. The interaction between people is complicated and depends on, for example, body language, general and facial appearance, speech, manners, clothing and age. The feeling of being different from others seemed to relate with several domains (Study I). Psychological problems have been reported to be associated with dentofacial appearance, speech difficulties and also a desire for further treatment (Ramstad, et al., 1995b; Turner, et al., 1998; Marcusson, et al., 2002). Good oral motor function and natural speech are important (Inglehart, 2002). Children and teenagers with CLP showed lower popularity cluster scores than a general young population (Leonard, et al., 1991). Accordingly, psychological support should always be available for persons with CLP and their families.

Regarding the facial appearance (question one, Study IV), both orthodontists and young adults without clefts identified “the teeth” as one of the first things they noticed on the photos of persons with BCLP. This emphasizes the importance of dental corrections. It is a subjective finding that is in line with the objective study of the frontal appearance of the dentition (Study III). A good symmetry of the upper frontal dentition and a straight midline between jaws were not always achieved but judged to be possible to improve in 40% of the cases. Considering the factors that influenced the ratings, Eliason et al. (1991) found that examiners who were familiar with CLP judged more negatively than those who were unfamiliar (Eliason, et al., 1991). However, in Study IV, the orthodontists were found to be less critical about the treatment results concerning the teeth of the persons with BCLP compared to the young adult evaluators. The reason could be that the orthodontists know the limits for what is possible to achieve with orthodontic treatment.

There were comments on poor oral hygiene, caries and gingivitis from both the professional and non-professional groups. Out of clinical experience, it is noted that many young adults with CLP develop varying degrees of aversion and tiredness related to their teeth and facial appearance. Persons who had lots of treatment are likely to be less interested in continuing treatment. In a large study on patients with severe disfigurements, either congenital or acquired, it was concluded that the satisfaction with their facial appearance would seldom reach the level of satisfaction experienced by non-disfigured persons (Versnel, et al., 2010).

To provide an optimum treatment outcome, frequency and timing of treatment, together with the quality of service, are of great interest. Combined orthodontic and surgical treatment is sometimes the solution for better results but many patients felt they already had enough treatment. Although, an Austrian study in 2005 presented that 63% of research participants asked for further treatment such as a lip-nose repair
(Sinko, et al., 2005). The results are probably highly dependent on how the questions are put. In Study III, only four of eleven candidates recommended for a combined surgical and orthodontic treatment accepted the offer. In Study IV, the judgments on the treatment outcome in terms of general appearance did not differ between the professional orthodontists and the young adults. They had similar opinions, both positive and negative aspects, including comments on details such as the form of the upper lip and the facial profile. This is in line with the logistic regression analysis. The statistics showed that each person’s individual opinion on each specific examination case was the most important factor, independent of the category of the evaluators (orthodontists or young adults).

All four studies are believed to be important in that new knowledge has been found regarding psychological matters, cleft morphology, other birth defects, dental appearance and orthodontic treatment results.
Conclusions

“Hoping to be like others” is the main category when persons with BCLP and iCP are interviewed using a “Grounded Theory” approach. The iCP does not affect the facial appearance which implies that this feeling is not only related to extra-oral looks.

One or more other birth defects can be seen in almost every third child with an iCP. A total iCP implies a higher risk for another birth defect compared to the risk when the iCP is partial.

A symmetrical upper front in young adults with BCLP seems difficult to achieve by orthodontic treatment. One reason is the high frequency of missing and/or peg-shaped lateral incisors.

A good sagittal relationship of the occlusion and a positive overjet can be achieved for a majority of young adults with BCLP.

The esthetic outcome after treatment of young adults with BCLP is evaluated differently on photos by a group of orthodontists and a group of young adults. The individual opinion is most important regardless of the category of evaluators. The group of young adults rated the esthetic outcome more negatively than the orthodontists.
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