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The Impact of Orofacial Clefts on Quality of Life and Health Care Use and Costs

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Abstract

Orofacial clefts are common birth defects that may impose a large burden on the health, quality of life, and socioeconomic well-being of affected individuals and families. They also result in significant healthcare use and costs. Understanding the impact of orofacial clefts on these outcomes is important for identifying unmet needs and developing public policies to reduce the burden of orofacial clefts at the individual, family and societal levels. This paper reviews and summarizes the main findings of recent studies that have evaluated the impact of orofacial clefts on these outcomes, with a focus on quality of life, socioeconomic outcomes, long-term health, and healthcare use and costs. Several studies identify an increased burden of orofacial clefts on these outcomes, but some of the findings are inconsistent. A summary of the primary limitations of the studies in this area is presented, along with recommendations and directions for future research.

Keywords

orofacial clefts; cleft lip; cleft palate; quality of life; healthcare costs; healthcare use

Introduction

Orofacial clefts (OFC) are one of the most common birth defects and occur in 1 per 500 to 2,500 births depending on ancestry, geographic residential location, maternal age and prenatal exposures, and socioeconomic status (SES) (Mossey and Little, 2002; Clark et al., 2003; Durning et al., 2007). Recent studies suggest that orofacial clefts are one of the most prevalent birth defects in the US, with more than 6,500 born in 2001 (CDC, 2006). OFC occur in three main types: cleft lip only (CL), cleft lip with palate (CLP), and cleft palate only (CP). More than 60% of cases with OFC have CL or CLP (CDC, 2006). The majority of cases with cleft lip with/without cleft palate (CL/P) are nonsyndromic (NS) (Jones, 1988; Marazita, 2002), occurring without recognized syndromes or other major birth defects and developmental disabilities (Murray, 2002).

Several treatments including surgery, speech therapy, dental care and psychological support are available for OFC. However, OFC impose a large psychosocial and economic burden on

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affected families families and society (Berk and Marazita, 2002), and associated with several health problems and complications early in life such as problems with feeding or ear infections (Nackashi et al., 2002), which can result in significant morbidity risks and also increased mortality risks, especially in less developed settings, where early systematic pediatric care may not be commonly accessible (Wehby et al, 2006a). OFC may also reduce quality of life throughout the life span. Several of the effects of OFC may extend through adulthood resulting in increased mortality and morbidity (Christensen and Mortensen, 2002; Christensen et al., 2004). Despite the prevalence of OFC, a paucity of information exists on the quality of life, long-term health and healthcare use and costs of affected individuals and families. Understanding the effects of OFC on the well-being of affected individuals and families and identifying healthcare needs is critical for making changes in healthcare practices and public policies to improve the health outcomes of affected individuals and families and for reducing the burden of OFC at the individual, family and societal levels.

The primary goal of this paper is to review the current research on health-related quality of life (HRQL), socioeconomic outcomes, health outcomes, and healthcare service utilization and costs. A secondary objective is to identify primary research gaps and potential study designs to address these gaps. This effort is complementary to other recent efforts, such as the Centers for Disease Control and Prevention (CDC) panel on identifying research priorities in OFC research (Yazdy et al., 2007).

Review of Health-Related Quality of Life Research on Individuals with Orofacial Clefts

A few studies on the effects of OFC on HRQL among children, adolescents and adults have been conducted. These studies have provided important preliminary insights into the relationship between OFC and HRQL. However, some of the study findings were inconsistent partly due to differences between studies with regards to patient populations, HRQL measures and study designs. Differences in HRQL measures were due to instruments employed, which ranged from HRQL instruments/questionnaires [such as the Pediatric Quality of Life Inventory (Varni et al., 2001; Damiano et al., 2007) or the Child's Perceptions Questionnaire (Jokovic et al., 2002, 2004; Wogelius et al. 2009)] to utility-based methods such as the visual analogue scales (Wehby et al., 2006b), as well as the groups whose preferences/perspectives for HRQL of OFC were measured, such as parents, patients, and health professionals. Studies also employed different designs, such as inclusion or exclusion of a control group of unaffected individuals. Common limitations of some studies were the reliance on small and convenient samples, which are primarily due to the challenges of identifying large and population-level samples of affected and unaffected individuals for such studies. Further, the majority of studies focused on assessing the effects of OFC on HRQL, but very few studies attempted to identify the factors that mediate the effects of OFC on HRQL. The most recent studies on HRQL among individuals with OFC are briefly summarized below.

Health-Related Quality of Life in Children with Orofacial Clefts

In the US, Damiano et al. (2007) measured maternal perceptions of child's HRQL in a sample of children 2–12 years old with OFC from Iowa. The authors found that HRQL decreased significantly with the presence of severe speech problems, and that older children with CP had lower HRQL compared to children with CLP. However, the study included no control group of unaffected children. Kramer et al. (2008) found no significant effects of the cleft type on child and parent-reported HRQL in a sample of children with OFC aged 5–6 years from Germany. The authors found that parental ratings of the child's HRQL were

lower than the child's ratings. The children's rating on the physical well-being domain decreased with the number of surgeries, but increased with the number of siblings.

Wogelius et al. (2009) found no significant differences in HRQL in a small sample of children with OFC and children without OFC aged 8–14 years from Denmark. Other studies found decreased quality of life of adolescents with congenital and acquired facial malformations compared to unaffected adolescents, as well as frequent reporting of stigmatization experiences (Topolski et al., 2005; Strauss et al., 2007). Quality of life decreased with the individuals' perceptions of increasing severity of facial malformations (Patrick et al., 2007). However, these studies included other congenital anomalies besides OFC.

Health-Related Quality of Life in Adults with Orofacial Clefts

Sinko et al. (2005) found lower HRQL among a sample of Chinese adults with repaired CLP aged 18–30 years, who wanted to receive more treatment compared to those who did not. Further, the study found that CLP impacted emotional and social functioning. Marcusson et al. (2001) evaluated the HRQL in a sample of adults with CLP and unaffected adults from Sweden. The authors reported significantly lower HRQL in the affected sample compared to the unaffected sample. The study found higher HRQL among adults who were more satisfied with their facial appearance (Marcusson et al., 2002).

Oosterkamp et al. (2007) evaluated the HRQL of a small sample of affected adults with bilateral CLP and unaffected adults from the Netherlands and found no significant differences in HRQL scores between the two groups, but higher HRQL among adults who were satisfied with their appearance.

Health Professionals' Perspective on Health-Related Quality of Life of Orofacial Clefts

Most studies of HRQL of OFC measured the preferences of affected individuals and parents. Variations in the preferences of patients, parents and health professionals for HRQL measurement were reported in previous studies (Saigal et al. 1999). Differences in the HRQL preferences of patients with craniofacial conditions and their parents have also been reported (Wilson-Genderson et al., 2007). Wehby et al. (2006b) measured the preferences of health professionals involved in providing craniofacial care to patients with OFC in the US for the overall impact of OFC on quality of life of affected individuals, using a visual analogue scale (VAS) method.[1] The study found that health professionals perceived a low effect of NS CL and NS CLP on HRQL, based on the VAS method. Study results suggested that professionals perceived a decreasing burden of OFC on HRQL with age of affected individuals, perhaps due to a larger emphasis on the surgical and medical treatments early in life than the long term health and psychosocial effects (Wehby et al., 2006b).

The study did not assess the preferences of patients or parents. However, given that the professionals', patients' and families' perceptions of the impact of OFC on HRQL may vary, a direct comparison by measuring the HRQL preferences of patients and parents using the VAS scale method becomes an important question for future research to address.

Impact of OFC on Socioeconomic and Psychosocial Outcomes

A few studies have assessed the effects of OFC on the socioeconomic and psychosocial outcomes of affected individuals and families. These studies have provided important insights into the psychosocial and socioeconomic burden of OFC. However, similar to the

[1]The HRQL scores were solicited on a scale between 0 for lowest quality of life possible and 1 for highest quality of life possible.

studies on HRQL, these studies were significantly limited by small and convenience samples and descriptive analyses that are subject to significant confounding factors, such as family-level and individual-level socioeconomic factors that may relate to both OFC and the studied outcomes.

Socioeconomic Outcomes

Kramer et al. (2007) did not find that OFC increased the financial burden of 130 families of children with OFC aged 6–24 months in Germany, but found that CP increased the family financial burden compared to CL or CLP. On the contrary, Kramer et al. (2008) found no differences in the financial impact of OFC by cleft type in another sample of families of 5–6 year old affected children.

In Norway, Ramstad et al. (1995) found overall no significant differences in employment and education of adults with CLP 20–35 years old compared to unaffected adults, but reported lower income, lower marriage rates, older age at marriage among the affected sample. Marcusson et al. (2001) reported a reduced economic performance among affected adults with OFC compared to unaffected adults.

Psychosocial Outcomes

Several studies related to the impact of OFC on the psychosocial status of affected individuals have been conducted. Several studies have reported psychological challenges among children, adolescents and young adults with OFC (Kapp-Simon et al., 1992; Kapp-Simon and McGuire, 1997; Hunt et al., 2006; Brand et al., 2009). Speech problems and concerns about esthetics are thought to contribute to these challenges (Thomas et al., 1997; Hunt et al., 2005; 2006; Patrick et al., 2007). Some studies have also found increased social anxiety among affected adults, though findings have varied between studies (Berk et al. 2001, Cheung et al., 2007). Christensen et al. (2004) reported a higher mortality rate due to suicide among individuals with OFC in Denmark compared to unaffected individuals.

Impact of Orofacial Clefts on Long-Term Health

Understanding the effects of OFC and other craniofacial conditions on long-term health outcomes is important for quantifying the health burden and improving service delivery and health care policies for affected populations. However, much remains unknown about the effects of OFC on individual and family long-term health outcomes and on healthcare needs. To date, only a few studies have examined long-term health outcomes, such as survival and occurrence of chronic diseases. One inherent limitation in conducting such studies has been limited access to appropriate data sources and health registries that track individuals with OFC throughout life and provide large population-level random samples of unaffected individuals.

Several studies using the Danish health registry system have assessed long term health outcomes of OFC. Christensen et al. (2004) found increased mortality risks for both males and females with OFC. Bille et al. (2005) found increased risks of breast and brain cancer among females with OFC and CP, respectively, and increased risk of lung cancer among males with CLP, all compared to unaffected individuals. Further, Christensen and Mortensen (2002) found significantly higher risks of hospital admission due mental health complications among adults with CP and CL/P, compared to unaffected adults. These studies strongly suggest that OFC imposes a large burden on the health of affected individuals throughout the life span.

Health Care Service Utilization and Costs of Children with Orofacial Clefts

In the US, several studies have examined health service utilization and costs among children with and without special health care needs using national datasets, such as the Healthcare Cost and Utilization Project (HCUP), Medical Expenditure Panel Survey, and the National Survey of Children with Special Healthcare Needs. The HCUP includes all payers of health services and is the largest collection of longitudinal health care data in the US (Russo and Elixhauser, 2007). However, these studies did not examine children with birth defects, including OFC, and did not verify the condition or diagnosis. Until recently, only two studies had been conducted on health service use and costs of children with birth defects, including OFC. This section summarizes the current studies on health service use and cost of children with OFC in the US.

Several cost perspectives exist, including the health care system, which are direct costs, and societal perspective, which includes all costs, for example caregiver costs and out-of-pocket expenses. Another viewpoint is the payer perspective, such as public and private health insurance, which measures costs with payments to providers. Depending on which perspective one is using, costs, charges, or expenditures is the appropriate measure of effect.

Because costs and expenditures are often used as a proxy for health care service utilization and are usually the most salient issues in terms of service delivery, program planning and policy development, this section focuses primarily on costs and expenditures of children with OFC and briefly mentions health service utilization. Differences in the results of these studies are attributable to using different: payers; definitions for healthcare service categories; ages; and units of analysis, such as hospital discharge (hospital stay) compared to an individual.

Studies conducted by Waitzman et al. (1992, 1994, and 1996) and Harris and James (1997) are the most comprehensive studies conducted on costs of birth defects, which included costs of OFC; however, they are now outdated. A major strength was the authors estimated costs from the healthcare and societal perspective and employed several major data sources to determine costs. Costs associated with mortality, morbidity, lost productivity, and developmental services and medical costs were included in the analysis. Despite the strengths of these studies, they did not provide information by cleft type or NS or syndromic status.

Several other studies have examined costs of children with OFC. However, these studies suffered from several severe limitations, including small sample size, selection bias, examination of charges, and not controlling for other confounding factors such as age and malformation type (Berk and Marazita, 2002; Snowden et al., 2003).

Four studies were recently conducted on hospitalizations and hospital costs of children with birth defects, which included OFC. Using the HCUP data, children of varying ages with OFC who had hospitalizations and costs for the year 2004 were examined[2]. The authors found that for children with NS CP, the total number of hospital stays was 2,900 per 100,000 affected children. In comparison, children with CL/P had a total of 4,900 hospital stays (Russo and Elixhauser, 2007). Russo and Elixhauser (2007) found that in 2004, the aggregate hospitalization cost for CL and CLP was about $11.6 million more compared to the aggregate costs of CP (Russo and Elixhauser, 2007).

[2]In this study, total hospital charges were converted to costs using HCUP cost-to-charge ratios. These were based on hospital accounting reports from the US Centers for Medicare and Medicaid Services (Russo and Elixhauser, 2007)

Using the HCUP 2003 Kids' Inpatient Database (KID) by the Agency for Healthcare Research and Quality, the authors found that in 2003, newborns with isolated CP had about 1300 fewer hospitalizations than newborns with CL or CLP (CDC, 2007). The 2003 HCUP KID study examined hospital costs during the newborn period and found the total hospital charges for CP were about $19 million more than the total hospital charges for CL and CLP, suggesting a higher cost per hospitalization for newborns with CP (CDC, 2007).

The major strength of these two studies was providing updated information on health care costs and charges of children with OFC. Yet, these studies suffered from several important limitations such as no stratification by presence of other anomalies, comparison to unaffected children, and the unit of analysis often was the hospital discharge, not the child.

The two most recent studies on health service use expenditures of children with OFC were from a public and private payer respectively. Primary strengths of these studies is that the authors: examined OFC by three different cleft categories and presence of other anomalies; examined different age groups; and compared these results to unaffected children (Cassell et al., 2008; Boulet et al., 2009). The study by Cassell et al. examined Medicaid health care use and expenditures in North Carolina during the first five years of life for children with and without OFC. The authors used a statewide, population-based birth defects registry to identify children with OFC born 1995–2002 continuously enrolled in Medicaid, which is the public insurance program for poor children and families in the US. The authors compared the results to a random sample of resident Medicaid-enrolled children without OFC born during the same time period. Using paid claims data, the authors examined several health care categories for service use and expenditures, including medical, inpatient, outpatient, home health, mental health, well-child care, dental and total. The authors found the total Medicaid expenditures for infants with OFC were higher than unaffected infants by about $11 million (Cassell et al., 2008). Mean expenditure of a child with syndromic OFC was almost five times that of a child with NS OFC (Cassell et al., 2008). In this study, total cumulative Medicaid expenditures over the first five years of life for children with OFC were significantly higher than unaffected children by about $22 million (Cassell et al., 2008).

A recent study examined health service use and insurer expenditures of children with and without OFC, ranging in age from birth to 10 years old (Boulet et al., 2009). The authors used the 2000–2004 MarketScan® Commercial Claims and Encounters databases, which provides data on health care use and expenditures of enrollees in several employer-sponsored plans. For children aged 0–10 years old with and without OFC, the difference in annual mean costs (incremental costs) was $13,405 (eight times higher than unaffected children). Mean costs for an infant with OFC and another major, unrelated defect were 25 times higher than those for an infant without OFC, and five times higher than for infants with an isolated cleft (Boulet et al., 2009).

Despite these studies' strengths, they suffered from several weaknesses. In both studies, the authors only examined direct costs and expenditures to the health care system. The study by Cassell et al. (2008) included crude expenditures and estimates may have not been representative of other states because Medicaid reimbursement rates vary between states in the US. These studies also did not include out-of-pocket expenses or caregiver costs (Cassell et al., 2008; Boulet et al., 2009).

Few data are available to identify determinants of variability in service use and costs, including patient (cleft severity, presence of other conditions), family (socioeconomic and demographic), and area-level (healthcare availability and access) characteristics. These factors are critical because patterns of medical and health-related service use and costs for

children with OFC can differ considerably by these factors. Additional studies should examine costs of outpatient care, dental care, speech therapy, and special education, with consistent estimations of out-of-pocket expenses, and caregiver costs to determine the true economic burden of OFC. Based in the US, the American Cleft Palate-Craniofacial Association, an international, multidisciplinary organization of healthcare professionals, has formed a Task Force to examine such issues. Currently, very limited information exists on the impact of OFC on indirect costs, such as loss in work productivity, time costs to parents, and effects of siblings' schooling. Understanding patterns of health service use and expenditures can help to target populations in need of services, assess the cost-effectiveness of treatments, and develop policies to improve the cost-effectiveness of and access to healthcare for patients and families with OFC.

Timeliness of and Referral to Services for Children with Orofacial Clefts

Services and treatment for children with OFC can vary depending on the cleft severity, presence of associated syndromes and/or other birth defects, and the child's age and needs (Nackashi et al., 2002). However, some general recommendations exist for services and treatment for children with craniofacial anomalies such as OFC (Nackashi et al., 2002; Lynch and Karnell, 2003; Canady et al., 1998; ACPA, 1993). These recommendations were originally set forth by the American Cleft Palate-Craniofacial Association (ACPA) in 1993 and were amended in 2000 and late 2004 (ACPA, 1993).

To date, only one study has examined the timeliness of such services in accordance with the ACPA recommendations (Cassell et al., In Press; Cassell and Meyer, 2008). This study was a retrospective study of North Carolina resident children with OFC born 1995–2002 who were continuously enrolled in Medicaid. The authors used North Carolina vital statistics, birth defects registry, and Medicaid enrollment and paid claims to examine the mean age at which surgery occurred and factors associated with timely cleft surgery among children with OFC. Using the 2000 ACPA guidelines, the authors found 78.1% of children with OFC had surgery within 18 months. The primary strengths of this study were using several data sources, including a birth defects registry, to provide information on the timeliness of services by maternal (age, race/ethnicity, education), child (cleft type and presence of other anomalies) and system (service type, residential location, receipt of maternity care coordination) factors associated with such services. The primary limitations of this study were the results were from one state and a public payer (Cassell et al., In Press; Cassell and Meyer, 2008).

One factor that can affect receipt of services, thereby affecting health service use and cost among children with OFC, is referral to services. Only three studies have examined referral to services among children with OFC (Cassell et al., 2007; White, 1981; Williams et al., 2003). These studies found that identification and referral to services of children with OFC, especially to craniofacial centers and teams, were significantly associated with location of residence, cleft type, presence of other birth defects, presence of other malformations in the family, and receipt of maternity care coordination services (Cassell et al., 2007; White, 1981; Williams et al., 2003). Due to the paucity of data on the identification and referral of children with OFC and other craniofacial anomalies to services, additional research is warranted to improve the timeliness of services and thereby quality of life and health outcomes for affected children.

Recommendations and Conclusions

The above sections summarized the findings and limitations of studies of the impact of OFC on the patient and family quality of life, socioeconomics, health outcomes, and health care use and costs. The primary limitations for conducting large-scale well-designed studies in

this area have been the limited access to and availability of large datasets that include measures of these outcomes for the OFC population and of comparison samples of unaffected individuals. Identifying large samples of affected individuals and the high cost of conducting large-scale prospective surveys to collect such data have been the main limitations. However, efforts to overcome these data shortages and expand the scale and scope of these studies is increasing.

A unique registry system that provides a strong venue for many of these studies, especially those focused on long-term health, healthcare use, and socioeconomic outcomes is the Danish Health Registry System (Christensen and Mortensen, 2002; Christensen et al., 2004; Bille et al., 2005). This data system includes a set of registries that include individual-level data on health care and prescription drug use, demographics, and socioeconomic characteristics for the entire population and allows for a random selection of large control samples, which provide an important methodological strength (Christensen and Mortensen, 2002; Christensen et al., 2004; Bille et al., 2005). This system has allowed for several important studies of long-term outcomes of OFC and is also enabling further ongoing studies to further identify the long-term effects of OFC on health, health care utilization, prescription drug and socioeconomic outcomes. The population-based data allow for consistent and generalizable estimations of such effects in large and representative samples.

A need exists for evaluating the impact of OFC on HRQL of affected individuals and families throughout the lifespan, using large population-based samples, robust HRQL measures, and from multiple perspectives, including the societal perspective. The societal perspective is needed for cost-effectiveness analyses of healthcare treatments for OFC (Gold, 1996). Further, it is important to employ both multi-domain survey instruments of HRQL, as well as methods that obtain HRQL values and utility scores, which are needed for cost-effectiveness analysis (Wehby et al., 2006b).

It is also extremely important to further understand the long-term effects of OFC on individual and family socioeconomic well-being. Some studies have identified increased risks of OFC with low socioeconomic status (Clark et al, 2003; Durning et al, 2007; Yang et al, 2008). Given that minimal changes occur in family socioeconomic status over time, baseline family socioeconomic status (prior to the birth of the child) may confound the assessment of the impact of OFC on long-term socioeconomic and financial outcomes. Therefore, studies investigating the impact of OFC on socioeconomic and financial outcomes should account for the family socioeconomic characteristics prior to the birth of the child. This also applies to studies of any outcomes that are influenced by baseline family socioeconomic characteristics, such as health outcomes, health care use and psychosocial outcomes.

Other important factors that should be accounted for in studies of OFC are maternal and parental preferences for risk taking and health. Maternal health behaviors such as smoking, alcohol use and multivitamin use affect OFC risks (Shi et al., 2007; Johnson and Little, 2008; Romitti et al., 1999). Health behaviors are a function of maternal preferences for health and risk taking and for child health, which are likely to influence other OFC related outcomes, such as healthcare use and costs. Therefore, it is important to account for these preferences, which may confound the relationship between OFC and quality of life, socioeconomic, psychosocial, health and healthcare outcomes. Measuring maternal and parental preferences for risk taking and health through assessment of relevant maternal health behaviors during pregnancy and accounting for these in studies of OFC outcomes is important for obtaining consistent estimates. Further, as the genetic risk factors of OFC are identified, genetic instrumental variable studies can be applied using these genetic variants as instruments for OFC when assessing its effects on quality of life, socioeconomic,

healthcare and other outcomes in order to account for unobserved confounders (Wehby et al., 2008).

Several important questions remain unanswered in terms of the effects of OFC on health care use and costs. A necessary understanding of health service use and costs of affected individuals over the lifespan is warranted. Estimating out-of-pocket, caregiver costs and indirect costs, including lost productivity, among affected individual and families using reliable methods and large scale datasets is needed. Also, a great need exists for examining the sources of variation in the type, quantity, and quality of health services provided to affected individuals and families and for estimating the cost-effectiveness of alternative treatment plans in order to identify ways to improve access of affected individuals to appropriate and cost-effective care. In addition, a need exists for further understanding of the role and effectiveness of integrated systems of care for individuals with OFC that involve craniofacial centers and teams, medical homes, dental, speech and mental health services. The role of health insurance in access to such systems is also an important component to examine further. Further research on the identification and healthcare referral patterns of children and adults with OFC and other craniofacial anomalies is warranted to improve referral to services, which can improve the timeliness of services and thereby quality of life and health outcomes of affected individuals.

In conclusion, several studies suggest an impact of OFC on the quality of life, socioeconomic and psychosocial well-being, long term health, health care use and costs for affected individuals and families. However, the primary limitation of most of these studies has been the reliance on small and unrepresentative samples with limited measures on several important outcomes and confounding variables. This in part has been due to the unavailability and lack of access to large-scale datasets that provide rich data for such questions. Therefore, a tremendous need exists for expanding the collaborations between various birth defect registries, craniofacial care providers, and researchers in order to identify data needs, improve data collection systems, and build consortia that provide access and opportunities to further examine the impact of OFC on multiple outcomes throughout the lifespan.

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