*Wednesday, February 7, 8:00AM-9:30AM*
*Concurrent Breakout Session E*

***Syndromes Associated with Congenital Heart Defects***
Moderator: Zenaida Steinhauer, New Jersey Department of Health & Senior Services, Trenton, NJ

Angela E. Lin, Massachusetts General Hospital, Boston, MA
Tiffany Riehle, National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention, Atlanta, GA

Congenital heart defects (CHDs), also known as cardiovascular malformations, occur as isolated birth defects in approximately 60-80% of affected individuals. In the 20-40% of individuals with a CHD and non-cardiac defect, a syndrome can be recognized in approximately 10-15%. Chromosome abnormality syndromes are more common than mendelian gene disorders.

The session presenters will review common as well as a few rare malformation syndromes and the associated CHDs.For each syndrome, the genetic basis, clinical and cardiac phenotype will be reviewed. Because this talk covers a broad spectrum of disorders and genetic topics, we will emphasize scope rather than in –depth discussion.

The combination of syndrome and CHD which will be highlighted are "*common syndromes with distinctive CHD pattern*", "*common syndrome with unremarkable CHD pattern*" and "*uncommon syndromes with distinctive CHD pattern*".

The syndromes will be categorized by genetic etiology including, 1) *chromosome abnormality* (e.g. trisomy 21/Down syndrome, trisomy 13 and 18, Turner syndrome, deletion 4p, deletion 7p/Williams syndrome; deletion 11q/Jacobsen syndrome, tetrasomy 22p/Cat-Eye syndrome, deletion 22q11 Spectrum/Velocardiofacial syndrome, DiGeorge syndrome); 2) *Mendelian gene disorders* (autosomal dominant, autosomal recessive, X-linked); 3) *known teratogens* (e.g. maternal diabetes, retinoic acid), and 4) *conditions with presumed but unproven genetic causation* (e.g. Adams-Oliver syndrome, Kabuki syndrome).

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***Health Services Utilization for Children with Craniofacial Malformations: Linkages with BD Surveillance Systems***
Moderator: Scott Grosse, National Center on Birth Defects and Developmental Disabilities, CDC, Atlanta, GA

**Health Service Use among Children with Orofacial Clefts in North Carolina**
Cynthia Cassell, North Carolina Birth Defects Monitoring Program, Raleigh, NC

***Background/Objectives:*** In North Carolina, about 180 children per year are diagnosed with orofacial clefts (OFC). For 1995-2002 in North Carolina, the prevalence of OFC was 15.29 per 10,000 live births. Orofacial clefts can impair teeth development, speech, hearing, feeding capabilities, and psychomotor and cognitive skills, creating emotional stress for infants, children, and their families. However, there is a paucity of current information on health care utilization and expenditures for children with OFC and other birth defects. National data that pertain to children with special health care needs (CSHCN) and selected subgroups of this population, such as children with asthma, fetal alcohol syndrome, cerebral palsy, and developmental disabilities are limited and do not specifically address children with OFC. Moreover, few data are available to describe variability in service use by maternal characteristics (e.g., age, race/ethnicity, and education) and child characteristics (e.g., age and diagnosis). These factors are critical because patterns of medical and health-related service use and expenditures for children with birth defects can differ considerably due to biological, familial, social, and developmental factors. The objective of this study was to examine patterns and predictors of health service utilization among children with OFC (cases) and a random sample of unaffected children (controls) in North Carolina. A secondary objective was to examine patterns of service use by cleft type and number of anomalies among the cases.

***Methods:*** Data from the North Carolina vital statistics, Health Services Information System, and North Carolina Birth Defects Monitoring Program were linked to identify resident children with OFC born between 1995 and 2002. Medicaid eligibility and paid claims were linked to identify resident children with OFC on Medicaid and a random sample of children on Medicaid born during the same time period and who were continuously enrolled (11+ months per year of life) in Medicaid during the first five years of life. Controls were selected in a 10:1 ratio and excluded any children with OFC or any other birth defects. Exclusion criteria for cases and controls included infant death within 12 months of life, out of state delivery and/or resident, and any adopted children. Univariate and bivariate analyses were conducted to determine patterns of service use by case-control status, cleft type and number of anomalies. Logit and ordinary least-squares regression analyses were conducted to examine predictors of service use among cases and controls for each year of life. Dependent variables were medical, dental, well-child care, outpatient, home health, inpatient, and mental health services. Independent variables consisted of selected maternal, infant / child, and system characteristics.

***Results:*** Among cases and controls, each category of service except well-child care for the first four years of life was statistically significant. Mental and home health services were utilized significantly higher among cases compared to controls. Overall, cases utilized health services almost twice as much compared to controls during the first five years of life. By cleft type, children with cleft palate (CP) and cleft lip with cleft palate (CLP) had similar patterns of service use compared to children with cleft lip (CL). In infancy, the mean number of Medicaid paid claims per child for medical service use for children with CP was 62.7 and 64.4 for children with CLP compared to 45.2 for children with CL. For all five years of life, mean total service use for children with CP and CLP were similar compared to children with CL. By number of anomalies, children with multiple anomalies (OFC and other birth defects) utilized services almost twice as much as children with an isolated anomaly (OFC only).

***Conclusions/Implications:*** This study provides information on service use by specific health service categories and by maternal, infant/child, and system characteristics for children with OFC compared to unaffected children. This study also stratifies health service use by cleft type and number of anomalies, which is important for allocation of resources, targeting care coordination and early intervention, etiologic research, and referral of services. As demonstrated, linking with several administrative data sources, population-based birth defects surveillance systems can play an integral role in examining health service use among children with OFC and other birth defects.

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***Health Services Utilization for Children with Craniofacial Malformations: Linkages with BD Surveillance Systems***
Moderator: Scott Grosse, National Center on Birth Defects and Developmental Disabilities, CDC, Atlanta, GA

**Children with Orofacial Clefts in Massachusetts: Hospital Utilization and Associated Costs**
*Marlene Anderka*, J Weiss, M Kotelchuck, D Wyszynski, S Grosse, R Garcia, W Barfield, H Cabral, E Lu, C Higgins, S Evans, S Manning, J Taubner

***Background:*** Orofacial clefts (cleft lip and/or cleft palate) are among the most common birth defects in the U.S. They are correctable defects that often require the intervention of a multidisciplinary team and multiple medical treatments to attain a satisfactory outcome. Thus, the financial burden of children with orofacial clefts on the health care system may be quite substantial. Related data on medical costs are sparse and outdated.

***Objective:*** To assess hospital utilization and associated costs in a population-based cohort of children born in Massachusetts with orofacial clefts

***Methods:*** This was a population-based cohort study using data from the Pregnancy to Early Life Longitudinal (PELL) Data System. Subjects included all children born in MA hospitals to residents (1998 through 2002) who were alive at age 2 years. Birth certificates were linked to hospital discharge birth records and post-birth hospitalization records by deterministic and probabilistic methods. Orofacial cleft cases were identified by the MA Birth Defects Monitoring Program. The comparison group was all other MA children without craniofacial malformations.

***Results:*** Over 70% of children with orofacial clefts born in MA during the 5 year study period had an isolated major defect with or without an associated minor defect. The median number of days children with isolated clefts were hospitalized was 3 times higher than in the comparison group (6 vs. 2 days). This difference doubled when children with non-isolated clefts were compared to the comparison group (12 vs. 2 days). The mean hospital cost from birth to age 2 among children with orofacial clefts was $21,090, and the incremental cost was $18,587. The average hospital costs for children with orofacial clefts were about 4 times higher during the birth hospital stay, 28 times higher for post-birth hospitalizations, and 8 times higher overall, compared to costs for all other children. Less than 40% of the mean total cost was attributed to the birth hospital stay among cases, versus 82% in the comparison group (mean total cost=$2,503). The total hospital expenditure during the first 2 years of life for children born with orofacial clefts in MA was approximately $10 million dollars (in 2003 dollars).

***Conclusions:*** In the first two years of life, children with orofacial clefts face increased hospitalization and incur increased hospital costs compared to children without craniofacial malformations.

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**Quality of Life and Craniofacial Anomalies: Initial Focus Group Results, Utah, 2006**
*Mary Bishop Stone,* Lorenzo D. Botto, Marcia L. Feldkamp, Ken Smith, LeAnn Roling, Duane Yamashiro

***Background and Objectives:*** Craniofacial anomalies affect approximately 4,000 newborns yearly in the United States. Although their clinical and genetic issues have been extensively studied, little is known about quality of life in infancy, childhood, and adolescence. As part of a comprehensive assessment of quality of life (which included also structured in-person interviews and mail surveys), we conducted two focus groups among parents of affected children.

***Methods:*** The two focus groups, one in Salt Lake City, Utah (12 participants), and one in Idaho Falls, Idaho (8 participants), included parents of affected children (10 boys, aged 5 months to 13 years; and 7 girls, aged 3 months to 18 years). The children had cleft lip and palate (n=14), cleft lip only (n=2) and cleft palate only (n=1). The focus group sessions lasted approximately 2 hours each, and were audio and video recorded and transcribed. Four investigators independently read the transcripts, and extracted domains. Then investigators as a group defined by consensus a list of common domains which were used to describe specific themes expressed by participants.

***Results:*** The following domains emerged from both focus groups as central concerns:
1. Insufficient information about diagnosis, management, outcomes: in particular, participants cited the scarcity of instructions for management after discharge (in particular regarding feeding); the lack of referral to specialists, including the craniofacial clinic, and support groups, such as other parents.
2. Insufficient preparation for surgery: in particular participants cited the lack of information about specialist, the lack of preparation for what the child would look like in the immediate postoperative period, the experience of mourning the way the child looked before surgery.
3. The experience of prenatal diagnosis: some participants would have preferred to know before delivery (to prepare themselves), others would have preferred not to know (to avoid worrying throughout pregnancy).
4. Costs and Insurance: many participants found the condition financially straining. Most insurance private insurances do not cover orthodontics which can be very expensive. Dealing with insurance companies was overwhelming for many, absent a parent advocate at the hospital or insurance company.
5. Social and peer issues: parents of older children emphasized the strain posed by teasing and staring at school and in public, because of facial appearance and speech challenges. School staff was sometimes helpful but not always.

***Conclusions:*** Several unmet concerns and needs emerged from the focus groups. Interventions aimed at improving health related quality of life associated with craniofacial anomalies may include the following:
- Strengthen communication with parents, from diagnosis throughout childhood and adolescence, using a variety of channels (phone, pamphlets, and websites) and realistic assessment of common concerns (feeding, daily life, and postoperative issues).
- Identify and publicize support structures, including parent groups and child advocates for dealing with hospital and insurance issues
- Improve school education to relieve teasing and peer pressure
- Facilitate ancillary treatment, including speech therapy.

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***Expanding the NBDPN Surveillance Guidelines: Data Presentation***
Moderator: Amy Case, Texas Department of State Health Services, Austin, TX

Amy Case, Texas Department of State Health Services, Austin, TX
Lowell Sever, Battelle Centers for Public Health Research and Evaluation, Seattle, WA

The NBDPN Guidelines for Conducting Birth Defects Surveillance is a resource that provides background information regarding surveillance and recommendations, with the goal of improving the quality and utility of surveillance programs and their data. The Surveillance Guidelines and Standards Committee has been working on a new chapter to provide information and guidance on presenting birth defects data. This new chapter will include consideration of the following topics: challenges and issues regarding data and factors influencing their presentation and interpretation, technical aspects of data presentation, graphics methods, geographic information systems (GIS), and privacy issues. After the introduction, Dr. Sever will review the rationale behind deciding what to include in the new chapter and will discuss the concepts and content being developed. The remainder of the time will be open for the audience to ask questions and provide feedback.