Distribution of Craniofacial Anomalies at Children's Hospital of New Orleans (CHNOLA) Craniofacial Clinic

Harrison D. Folse; Regina Zambrano, M.D.

LSU School of Medicine, New Orleans, Louisiana; Louisiana State University Health Science Center-Department of Pediatrics, Division of Genetics, New Orleans Louisiana and CHNOLA

Introduction

Craniofacial anomalies are a diverse group of congenital disorders, affecting a great number of patients around the world. An example, Cleft lip and palate (CLP) affects 1/500-700 births worldwide (Data from WHO). In the US, the average prevalence of cleft lip and palate is 10.63/10,000 live births. (Data from CDC). Craniofacial anomalies can be polygenic/multifactorial (80%) or syndromic (20%).

We are particularly interested in syndromic craniofacial anomalies and their management.

Our primary goal is to determine the prevalence of syndromic craniofacial anomalies in the population of patients attending the Craniofacial Clinic at The Children's Hospital of New Orleans (CHNOLA). In the year 2019, we recorded 570 visits to the CFS clinic,

Methods

We will conduct a retrospective chart review of patients seen in the Craniofacial clinic at CHNOLA from January 2010 to June 2020. We will use data from the REDcap database and electronic medical records (Epic and VCO). Patients seen in the clinic who have been diagnosed with a craniofacial anomaly will be included in the study.

The following information will be collected during the retrospective chart review:

1. Type of craniofacial anomaly
    a. Orofacial cleft or VPI
    b. Craniofacial microsomia
    c. Craniosynostosis
    d. Dental anomalies
    e. Other
2. Etiology of Craniofacial anomaly
    a. Syndromic
    b. Non syndromic (Isolated/Polygenic/Multifactorial)
3. Gender
4. Age
5. Ethnic background

Actions and Plans

For each patient, we will first determine whether their craniofacial anomaly is syndromic or polygenic/multifactorial. If it is polygenic/multifactorial, we will make sure the patient/family has been counseled on the future outcomes. If the patient's craniofacial anomaly is syndromic, we will follow a different protocol. Along with counseling to make sure the patient/family is aware of any chances of recurrence, we determine if the molecular etiology for the craniofacial anomaly has been identified; if not, we will work on finding the etiology. For the syndromic patients, we will also create "roadmaps", which will be included in the patient's chart so all members of the healthcare team will know the appropriate treatment for the patient.

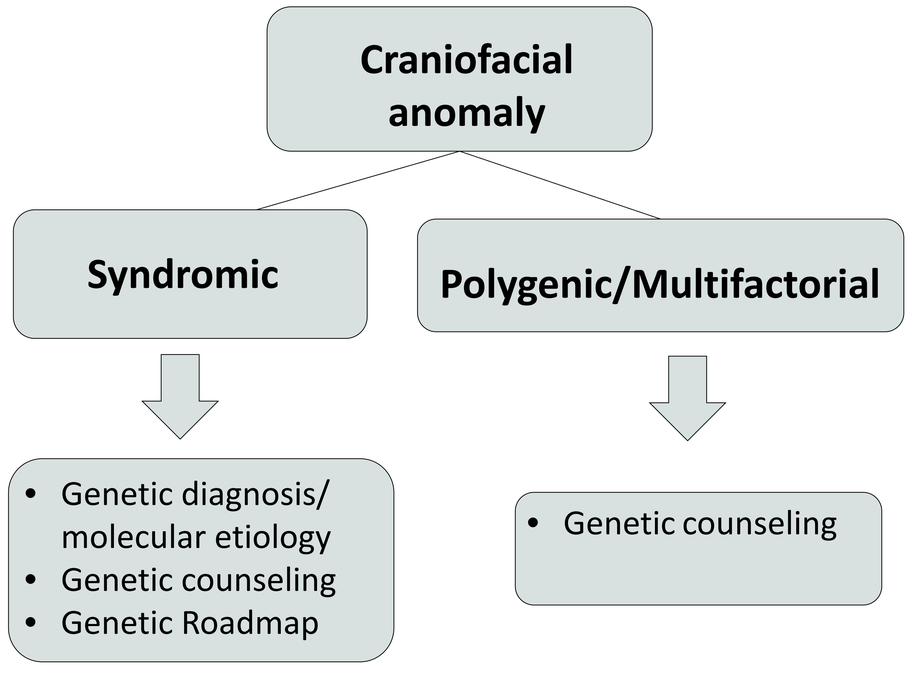

Figure 1.

Future Directions

Based on this information, we will then create "roadmaps" of what to do for a patient's specific diagnosis. These "roadmaps" will be placed in the patient's chart and be used as a guide for their management.

This will allow any member of their healthcare team to familiarize themselves with rare disorders, identify the patient's needs and provide adequate care.

We are hoping to offer a more organized and comprehensive care for our families in the CF clinic at CHNOLA. This will improve and increase access to treatment for patients, while allowing the healthcare team to have a better understanding of their patients' needs.

Sample Genetic Roadmap

22q11 Genetic Roadmap

Patient Name: _____ Patient DOB: _____ MRN: _____

Assessment	At diagnosis	0-12 months	1-5 years	6-11 years	12-18 years	>18 years
Genetic confirmation (22q11 FISH)						
Thyroid function testing						
Ionized Calcium, PTH						
CBC and differential						
Immunology evaluation						
Ophthalmology evaluation						
Palate evaluation						
Cervical spine evaluation						
Audiology evaluation						
Scoliosis evaluation						
Dental evaluation						

Children's Hospital New Orleans LCMC Health

Assessment	At diagnosis	0-12 months	1-5 years	6-11 years	12-18 years	>18 years
Renal US						
Echocardiogram						
EKG						
Developmental assessment						
Psychiatric evaluation						
Parental studies						
Genetic counseling						
Gynecologic assessment						

Children's Hospital New Orleans LCMC Health