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“Distribution of Craniofacial Anomalies at Children’s Hospital Craniofacial Clinic and Plans for Genetic Roadmaps for Management”

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.

The intent of this study is to determine the prevalence of syndromic craniofacial anomalies in the population of patients attending the Craniofacial Clinic at The Children’s Hospital in New Orleans (CHNOLA).

We will conduct a chart review of the patients that attended/will attend the Craniofacial Clinic at CHNOLA. We will include all patients seen by the LSUHSC Genetics department for craniofacial anomalies (including cleft lip, cleft palate, cleft lip and palate CLP, craniosynostosis, hemifacial microsomia and other anomalies) from July 2010 to the present.

The project will look at the following variables:

1. Is the anomaly isolated or syndromic?
2. If syndromic, has the molecular etiology been identified?
3. Has the family received appropriate counseling regarding the chances of recurrence for isolated/multifactorial and syndromic cases?

This data will then be compared to national published data. 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.