

*Tuesday, February 24, 8:00AM-12:00PM
Plenary Session*

Here's Looking at You: Defects of the Head and Neck

Moderator: Angela Lin, Massachusetts General Hospital, Boston, MA

The Pathogenesis and Etiology of Cleft Lip and Cleft Palate: One Clinician's Journey

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Facial clefts may be understood either from the perspective of what went wrong in embryogenesis (pathogenesis) or from the perspective of what caused the defect (etiology). In some instances we understand one or the other. In other instances we understand both.

This presentation will review the results of the author's almost 30 year experience in the evaluation of over 2000 consecutive cases of cleft lip with or without cleft palate and cleft palate alone, classifying the conditions both by pathogenesis and etiology. Several conclusions may be drawn:

- 1) most clefts represent malformations in development although deformation, disruption and dysplasia also account for a minority of cases;
- 2) syndromes account for roughly 15% of cases of cleft lip with or without cleft palate and 40% of cases of cleft palate alone;
- 3) a few syndromes account for the majority of the "syndromic" cases;
- 4) mutations in genes responsible for "syndromic" forms of clefting may also confer susceptibility to non-syndromic clefting;
- 5) phenotyping both respect to etiology and pathogenesis is critical to the identification of genes that confer susceptibility.