Heart defects in babies with cleft lip and palate linked to gene defect

The Centers for Disease Control and Prevention (CDC) report that approximately 2,650 babies are born with a cleft palate every year, and another 4,440 with a cleft lip.

While these orofacial birth defects are common, their cause remains largely unknown. Genetic changes as a result of the mother’s diet or medication are thought to play a role, but the exact genetic mechanism is not fully understood.

A team of international researchers from the United Kingdom, Canada, Saudi Arabia, and the U.S. has set out to investigate the genetic mutations behind cleft lip and palate (CLP) and their accompanying heart defects.

Using genetic mapping, the researchers looked for the chromosomal location of the gene responsible for the disease…After adjusting for various factors, the team found only one pathogenic variant in the HYAL2 gene[, which encodes an enzyme responsible for degrading hyaluronan.]

Given that hyaluronan is found in the connective tissue of many parts in the body, including the heart, the researchers hypothesized that mutations in the HYAL2 gene would cause CLP and heart defects in mice.

The GLP aggregated and excerpted this blog/article to reflect the diversity of news, opinion, and analysis. Read full, original post: Scientists find genetic mutation that causes cleft lip and palate, heart defects