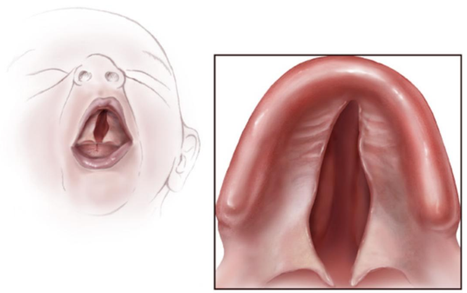
Mouse models of human cleft lip and/or palate

Charles W. Griffin[1], Thomas E. Forman[1], and Katherine A. Fantauzzo[1,2]

[1]Department of Craniofacial Biology, University of Colorado Anschutz Medical Campus, Aurora, CO, USA; [2]RNA Bioscience Initiative, University of Colorado Anschutz Medical Campus, Aurora, CO, USA

How common is cleft lip and palate and what is it?

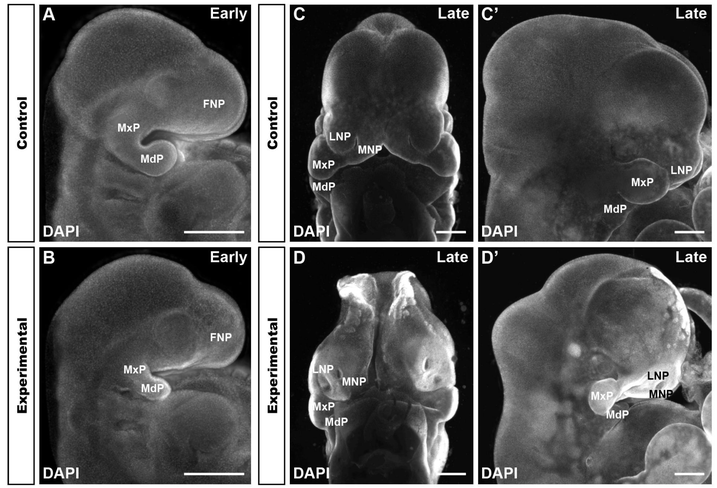


Centers for Disease Control and Prevention, National Center on Birth Defects and Developmental Disabilities

- 1 in every 1,050 babies born with cleft lip with or without cleft palate
- Cleft: a split between the right and left sides of the face
- Palate: the roof of the mouth
- Children with cleft lip or a cleft palate usually require surgery as they may have problems with eating and breathing, and as they grow older may have speech and language delays
- These children are also more prone to have ear infections, hearing loss, and problems with their teeth

National Institute of Dental and Craniofacial Research

Underdeveloped face and facial clefting in mice

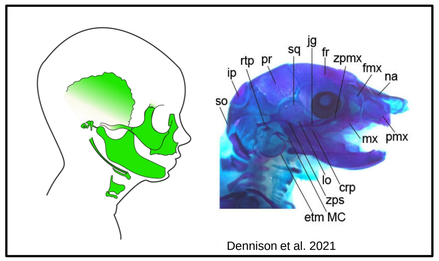


Major Takeaways

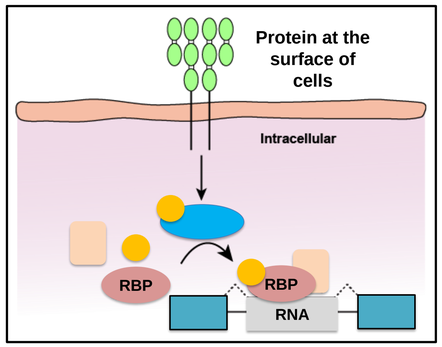
- Cleft lip and/or palate is a common facial difference in humans.
- RNA-binding proteins play a role in how our face develops.
- Disruption of one of these RNA-binding proteins results in an underdeveloped face and severe facial clefting.
- This research provides insight into the requirement for proteins that interact with RNA during the development of the face.

How proteins are important for the development of the face

- Migration of the cells that make up the face give rise to the bones and cartilage of the facial skeleton
- This process is the same between humans and mice, allowing us to use the mouse model to explore the mechanisms underlying how the face develops and disruptions to this process in the context of human disease

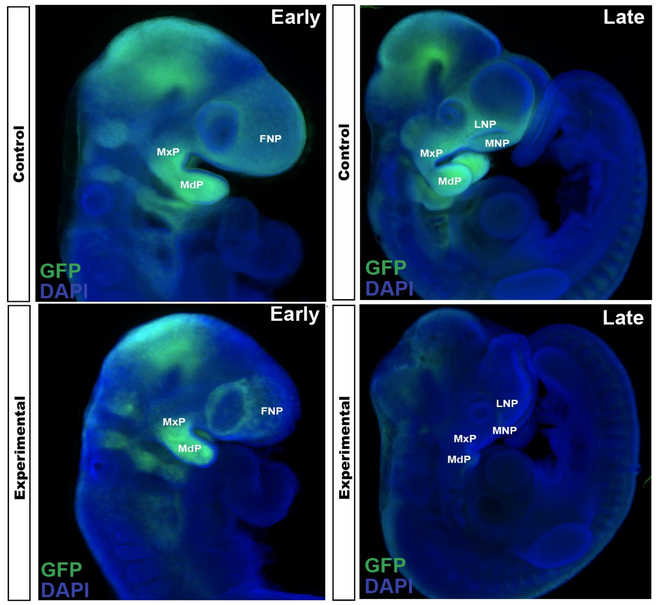


Dennison et al. 2021



Signaling through proteins at the surface of cells can change how proteins inside the cell bind to RNA, a precursor of protein. These RNA-binding proteins are necessary for the development of the face.

Defective growth and survival of cells that make up the face



Future Directions

Continue exploring how RNA-binding proteins affect the development of the face

Investigate what other proteins these RNA-binding proteins interact with

Test for mutations of these RNA-binding proteins to identify potential genetic conditions early in human development

BioRender

ACKNOWLEDGEMENTS

We are grateful to Erin Binne for technical assistance. This work is supported by NIDCR grants R01DE030864 (to K.A.F.) and F31DE032252 (to T.E.F.).