Whole Exome Sequencing Identifies a Novel Candidate Gene in an Ashkenazi Jewish Family with Tetralogy of Fallot

JAR-YEE LIU*, AFRIDA SARA*, Jason Liu, Pritha Gupta, Judith Fan, Jessica Wang
UCLA Department of Medicine - Division of Cardiology
David Geffen School of Medicine at UCLA

Background

Figure 1: Diagrammatic Depiction of Tetralogy of Fallot (ToF). 70% of ToF Patients Lack of a Molecular Diagnosis

Precision Medicine

Figure 2: Precision Medicine. Delivering the right treatment, to the right person, at the right time. Removes delay, expense, and harm of trying ineffective treatments

Whole Exome Sequencing (WES)

Figure 3: Whole Exome Sequencing & Analysis Workflow

Methods & Tertiary Analysis

Figure 4: Familial Pedigree. Two affected sisters (red) with no pertinent family history.

Discussion

Figure 5: Cardiac catheter visualization of proband’s 48-year-old sister.

Figure 7: Real Time PCR Analysis for DSPP in non-mineralized mouse tissue

Future Directions

- Immunohistochemistry for DSPP visualization in developing embryonic mice
- Molecular cloning for gene expression in cell culture

Figure 8: β-Galactosidase Assays for DSPP in non-mineralized mouse tissue

Acknowledgements

Figure 9: Odontogenesis.