



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

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Background

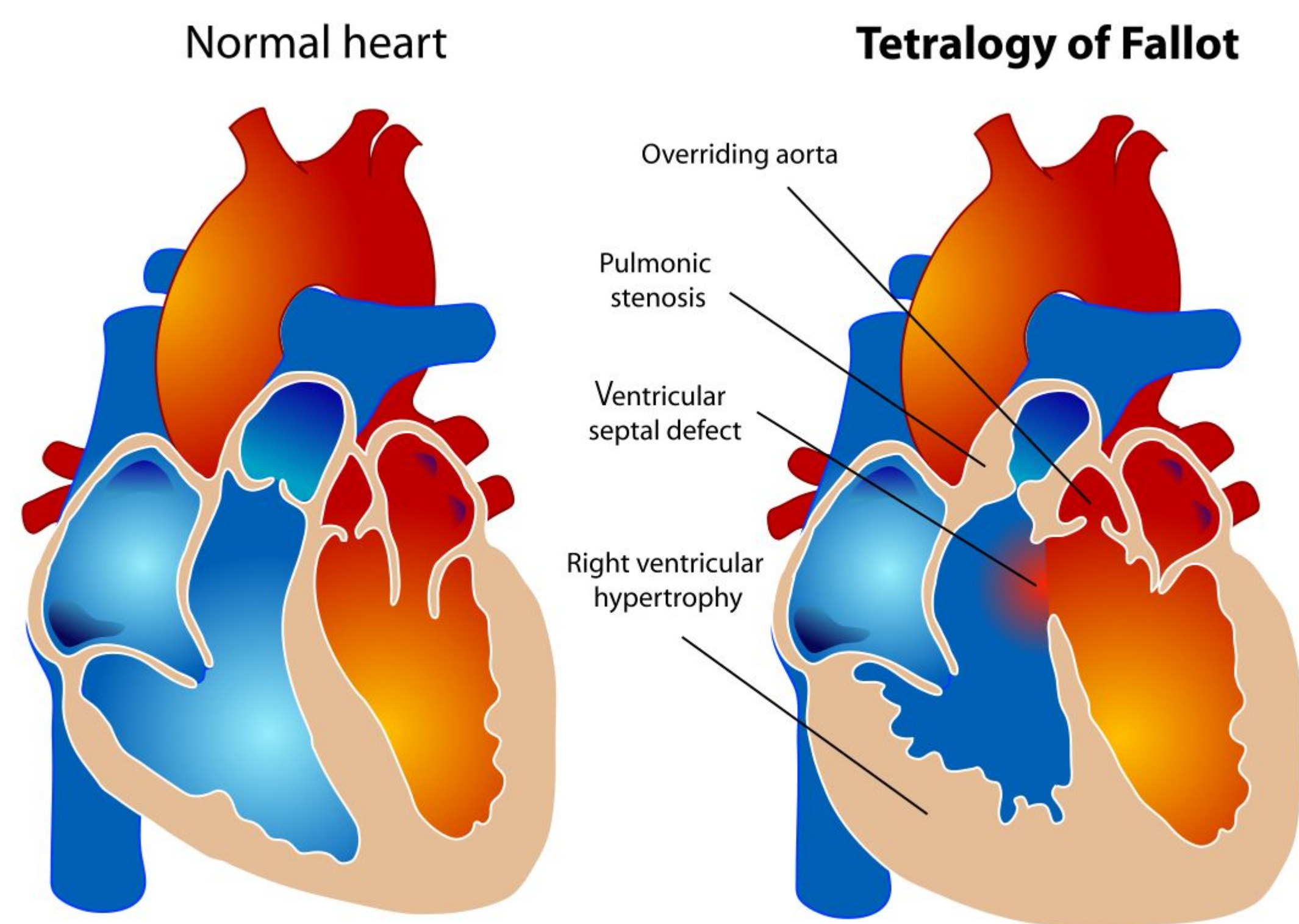


Figure 1: Diagrammatic Depiction of Tetralogy of Fallot (ToF). 70% of ToF Patients Lack of a Molecular Diagnosis

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)

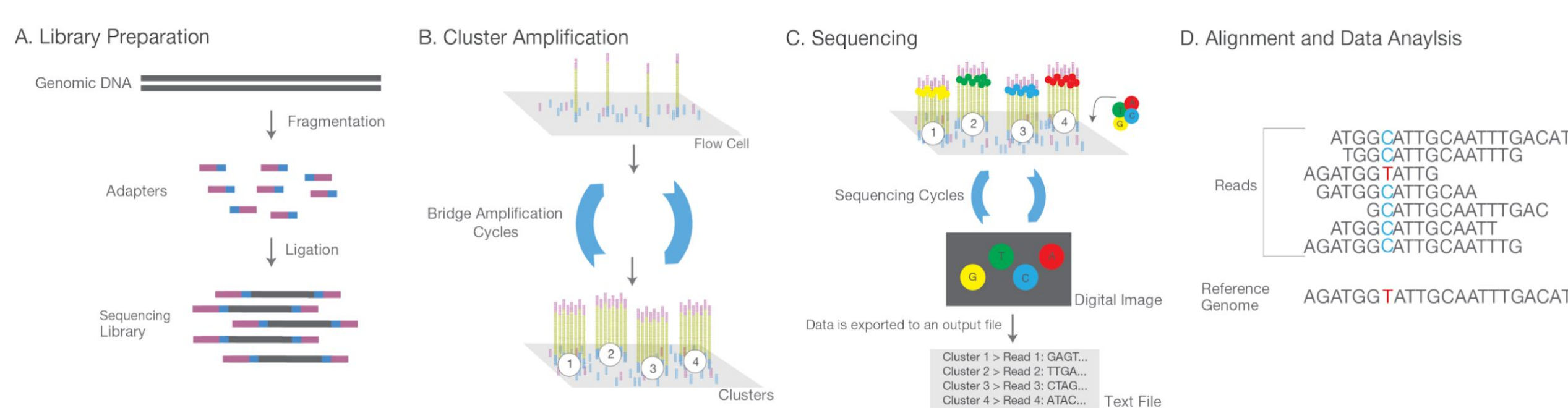


Figure 3: Whole Exome Sequencing & Analysis Workflow

Goal

To identify a genetic basis for development of Tetralogy of Fallot (ToF) within an affected family

Methods & Tertiary Analysis

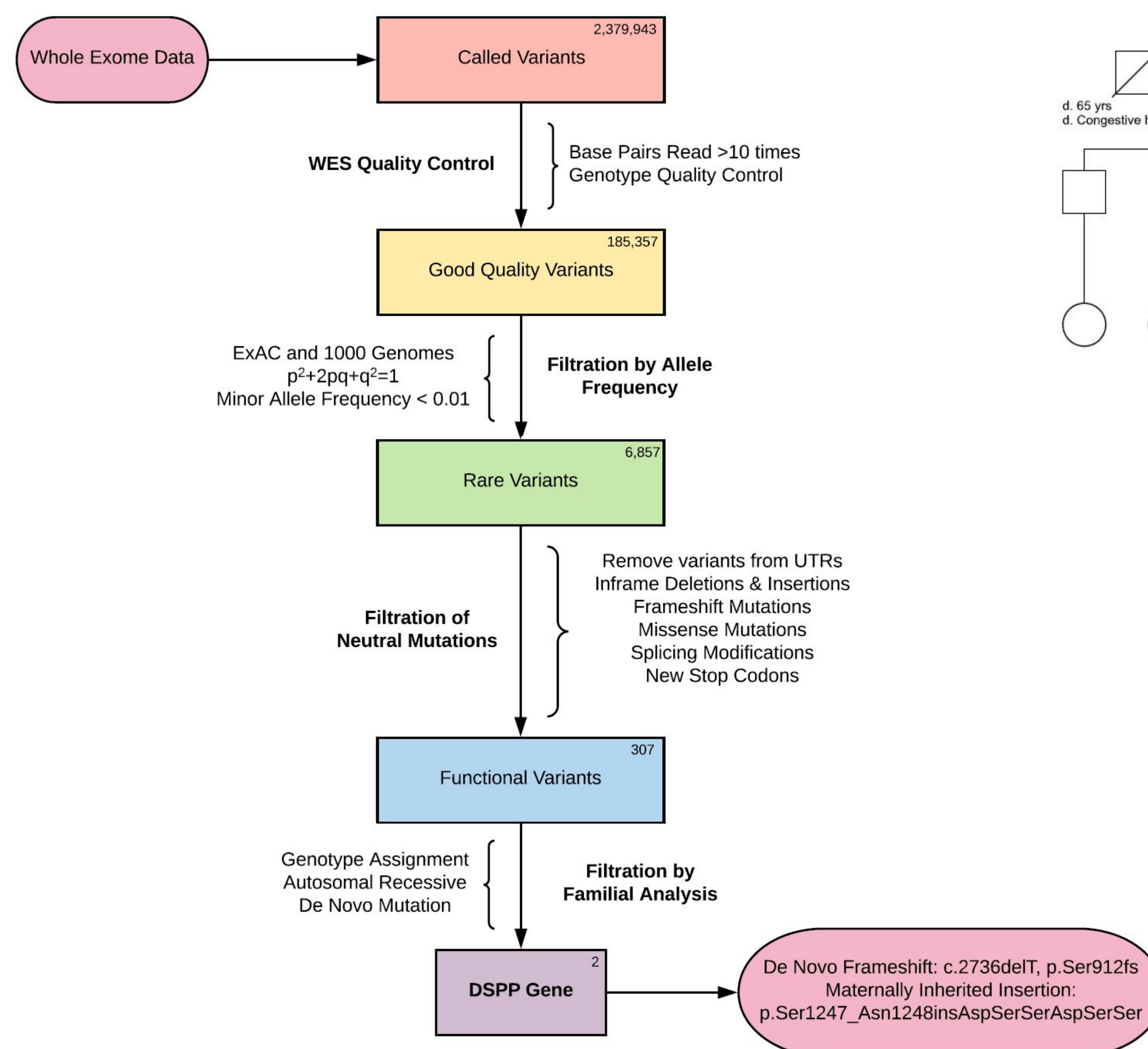


Figure 6: Computational Exome Analysis Workflow

Discussion

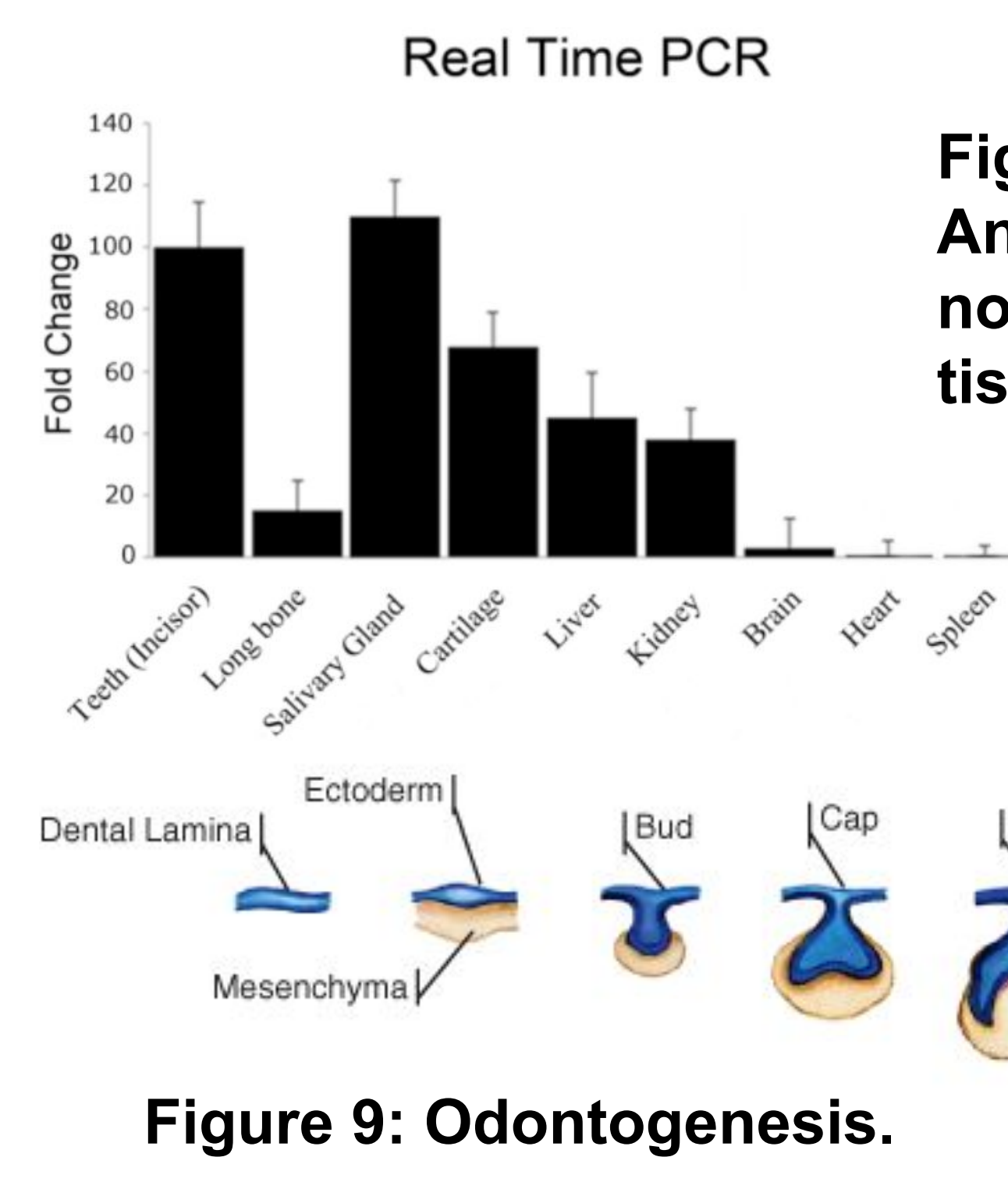


Figure 9: Odontogenesis.

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

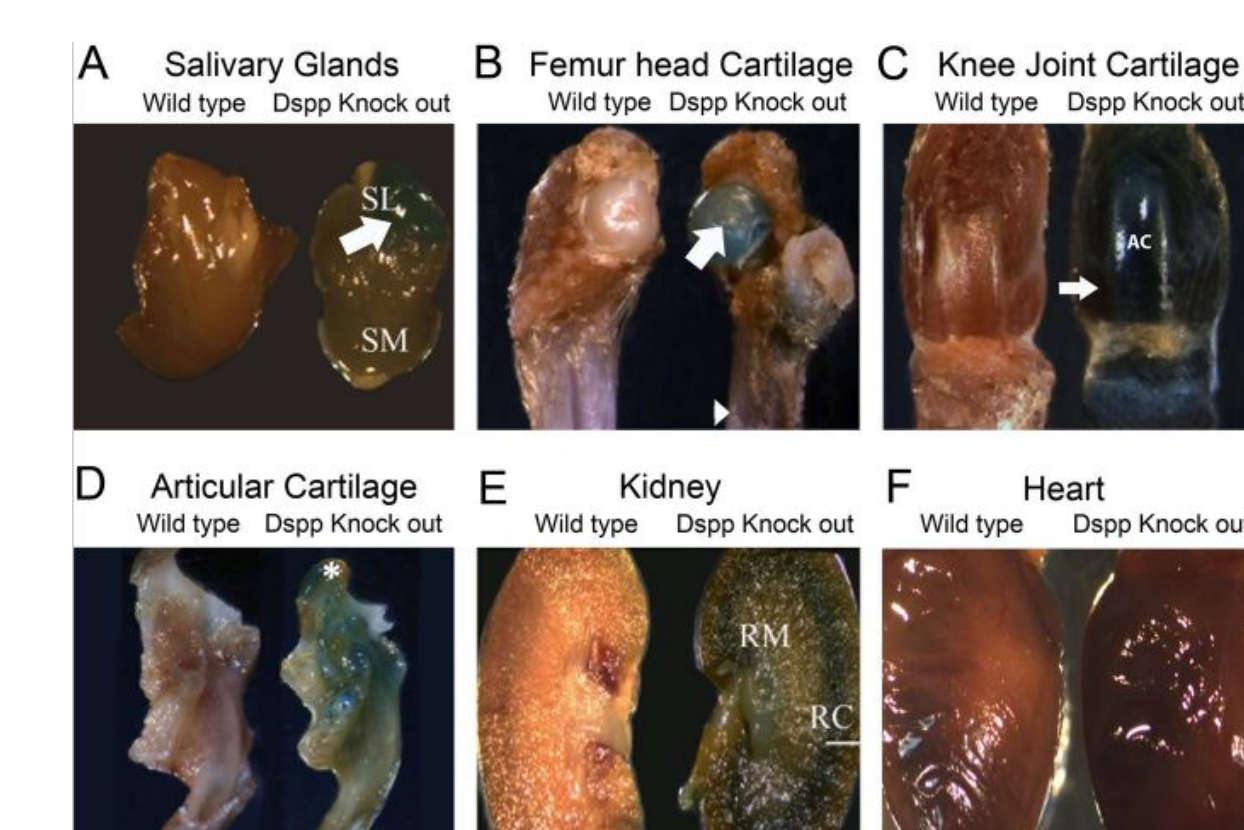


Figure 8: β -Galactosidase Assays for DSPP in non-mineralized mouse tissue

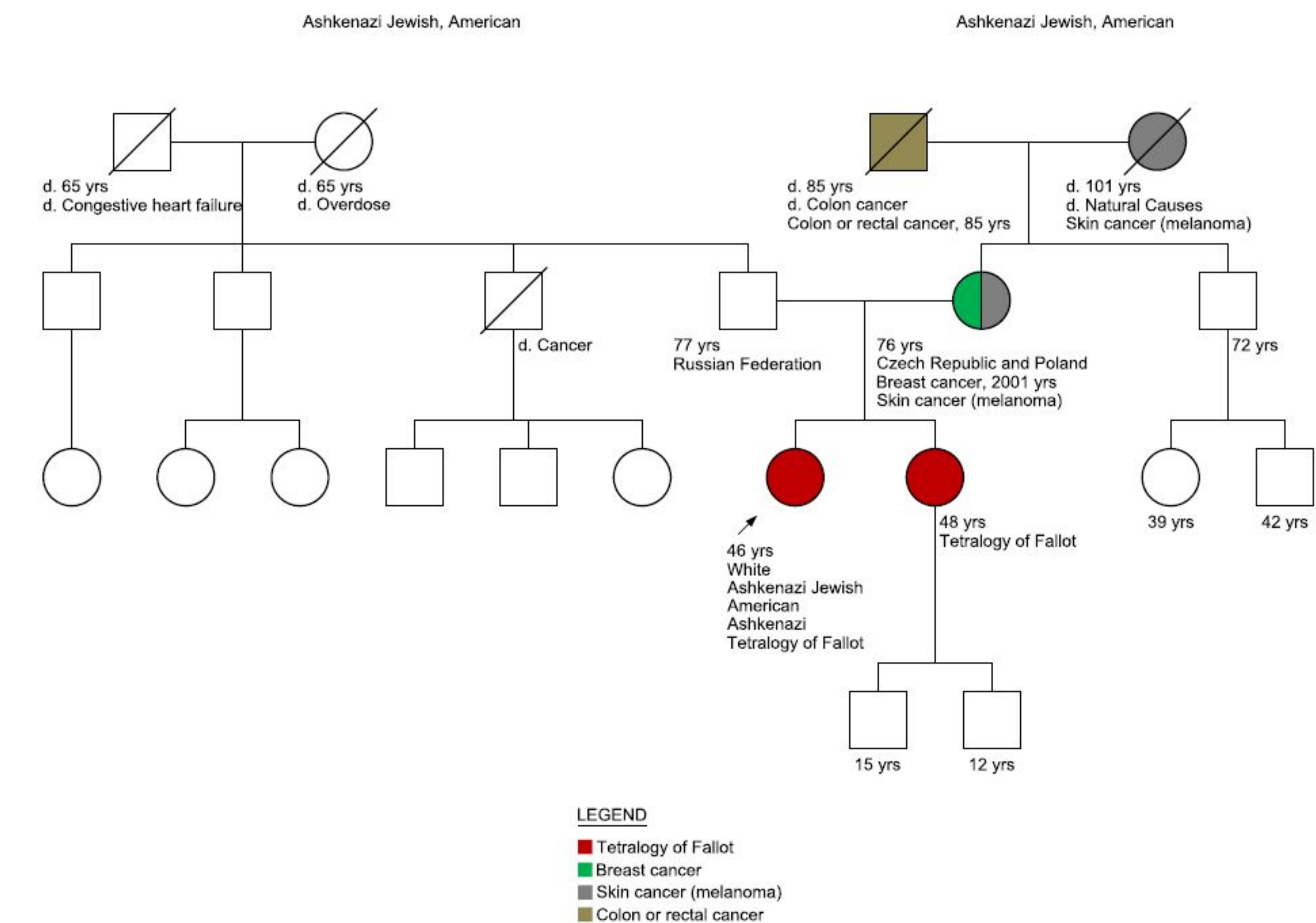


Figure 4: Familial Pedigree. Two affected sisters (red) with no pertinent family history.

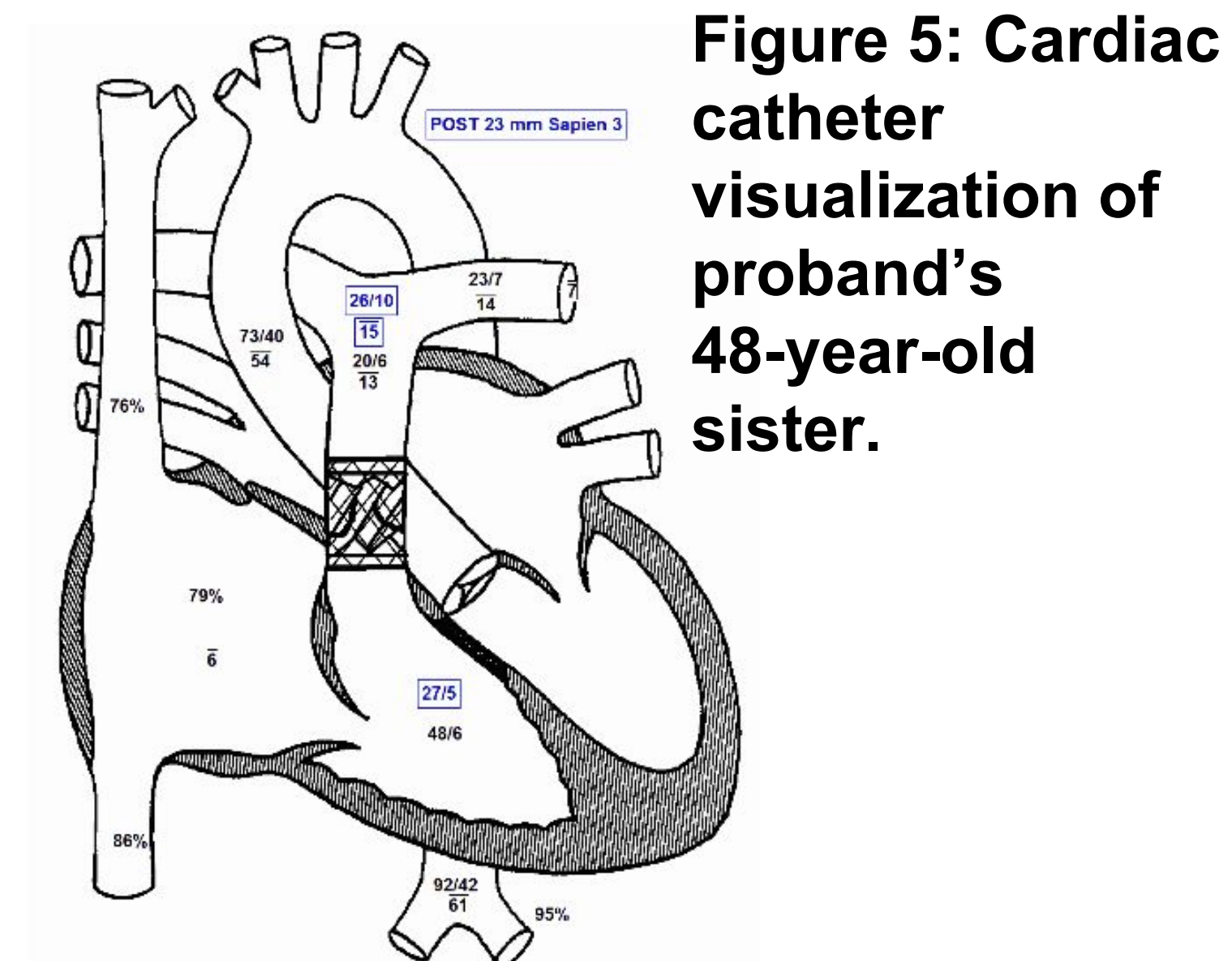


Figure 5: Cardiac catheter visualization of proband's 48-year-old sister.

Future Directions

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

Acknowledgements

