## Introduction:

- Ellis van-Creveld Syndrome (EVC) is a rare condition that is characterized by short limbs, polydactyly, nail dysplasia, and other developmental afflictions.\(^1\)
- Mutations in EVC and EVC2 genes lead to the development of Evc.\(^1,2\)
- EVC and EVC2 proteins are positive modulators of the Indian Hedgehog signaling pathway which regulates the growth of bones and related tissue.\(^3,4\)
- The two proteins interact with each other and localize on the primary cilia membrane.\(^4\)
- The precise role of the proteins in the Indian Hedgehog signaling pathway has not yet been determined.

## Strategy to generate complete EVC gene:

**Figure 1.** Generation and cloning of the full-length hEVC gene from human heart cDNA library, including (A) overlapping fragments, (B) fragment sizes, (C) overlap extension PCR, and (D) ligation into vectors.

## Future Work:

- Confirm DNA sequence of human EVC gene product
- Optimize expression protocol
- Characterize biochemical properties of hEVC protein

## Acknowledgements:

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## Results:

**Figure 2.** Isolation of Fragments 2 and 3, 787 bp and 777 bp respectively, from human heart cDNA library via PCR.

**Figure 3.** Generation of full-length hEVC gene from three fragments. (A) Fragments 2 and 3, 787 bp and 777 bp respectively, were ligated to form Fragment 4, with a length of 1467 bp, via SOE-PCR. (B) Fragments 1 and 4, 1537 bp and 1467 bp respectively, were ligated to form the full-length hEVC gene, with a length of 2976 bp, via SOE-PCR.

**Figure 4.** Cloning and expression of full-length hEVC gene, involving (A) pGEMT-Easy vector map, (B) confirmation of construct via double digestion, (C) time scale expression analysis with SDS-PAGE, and (D) p202 vector map.

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### References:


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