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:

Future Work:

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

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References: