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Greetings!

We are presently undertaking a genetic study to detect the chromosomes and genes causing Goldenhar syndrome (also known as hemifacial microsomia or oculo auriculo vertebral dysplasia). Genes are the units of information we inherit from our parents and which guide the development of a fetus. The genes are then organized into structures which are present in all cells of our body. The genes involved in Goldenhar syndrome have been changed so that it alters development of the face, ears, eyes and other organs.

We anticipate that this study to isolate the genes involved in Goldenhar syndrome will take time to complete, and it will only be accomplished if we can enlist the help of individuals who have this condition and their families. Our results will be useful as we seek to understand how this condition occurs. Isolating the genes will also result in the availability of prenatal diagnosis for families for who may one day be interested in this service.

We are excited about this study and hope that you and your family will choose to participate. In order to do so you will need to do the following: sign a Johns Hopkins Medical Institutions clinical consent form, fill out a questionnaire about your medical and family histories and provide a small blood sample (2 tablespoons). Individuals with Goldenhar syndrome and, in some cases, unaffected relatives, especially parents, may participate. In this way, we hope to study the chromosomes and genes from the affected individuals as compared to their unaffected relatives. Of course, there is no charge for your participation, and we will keep you informed of our progress. Information pertaining to you will be held confidential.

If you have any questions or if you want to participate, please do not hesitate to contact me through my email at ejabs1@jhem.jhmi.edu or through our address on the letterhead.

Yours Sincerely,

Dr. Ethylin Wang Jabs, MD
Director, Center for Craniofacial Disorders