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Genetic linkage analysis and candidate gene approach in Puerto Rican families with Congenital Scoliosis

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Scoliosis can be either idiopathic or secondary to congenital vertebral abnormalities. Idiopathic typically occurs in children and adolescents who are otherwise healthy. Congenital is due to anomalous development of the vertebrae (failure of formation and/or segmentation). Incidence is estimated in 1/1,000 to 1/2,000 but the true incidence remains unknown. Etiology is unknown. Age and range mean 6 years (range few days of life to 25 years). Is more common in girls than in boys, occurring in the ratio of 2.5 - 1. Genes of the Notch signaling pathways have been identified as responsible for some cases of scoliosis, they are involved in various biologic processes, including somatogenesis, and neurogenesis. Our primary objective will be to identify genetic component in congenital scoliosis using family linkage (consanguineous families) analysis and association studies that will help in the identification of the genetic component or disease susceptibility to a specific chromosomal region that may help in unraveling of the underlying defect. Understanding and defining the natural history of specific mutation(s) and the developmental (molecular) mechanisms in vertebral patterning, may aid in the identification of protective factors for normal spinal development and toward the prevention of disfiguring congenital scoliosis. No study of this nature has ever been conducted in the Puerto Rican population and very limited data of incidence and prevalence ever been reported.

Biography

Juan E. Baez-Segarra is a doctoral candidate in Public Health at the Ponce School of Medicine and Health Sciences, Ponce, Puerto Rico. He has a DVM degree from the Pedro Enriquez Ureña University in Santo Domingo, Dominican Republican. He has been practicing for 20 years as a small animals veterinarian. Dr. Baez is also an advocate for the local Animal Shelter. He possess a master's degree in Public Health from Ponce School of Medicine.

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