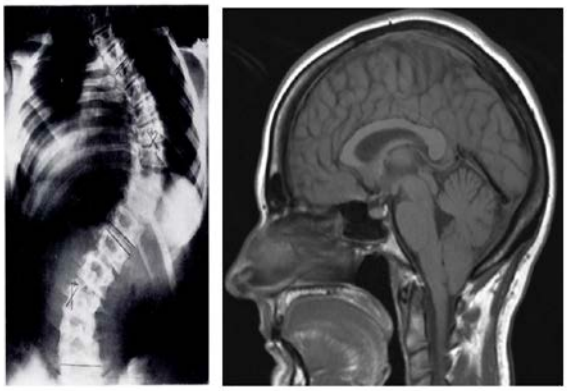
GUEST SPEAKER SEMINAR

Molecular Genetics
UNIVERSITY OF TORONTO

Are there overlapping pathogenetic mechanisms chiari 1 malformation and scoliosis?



Chiari Malformation Type 1 (CM1) affects 1 in 1000 individuals symptomatically though and may be observed radiologically in up to 1%-3.6% of MRIs, making it a common disorder that represents a substantial personal, familial and societal burden. Many CM1 patients develop syringomyelia, a fluid filled cyst in the spinal cord resulting in motor and sensory deficits and urinary incontinence. Interestingly, 20% of CM1 patients develop scoliosis, although this figure increases to 60% when syringomyelia is present. The pathogenetic mechanism by which patients with CM1 develop syringomyelia and scoliosis are not fully known. Our group recently evaluated the underlying etiology and comorbidities of more than 600 CM1 cases which revealed heterogeneous causes, with only 5% having a known genetic disorder, with overgrowth syndromes and Ehlers-Danlos syndrome making up the largest proportion. We are now undertaking a large-scale whole exome sequencing study of more than 1000 adolescent idiopathic scoliosis cases and 500 CM1 cases which will allow us to determine whether there are overlapping genetic risk factors associated with these related conditions. Generating animal models to allow us to more completely understanding the pathogenetic mechanisms will be essential for developing new preventive strategies for these severe disorders.

Dr. Christina Gurnett, MD PhD

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Host: Dr. Brian Ciruna

Date: Monday, September 23rd
Time: 10AM
Place: Terrence Donnelly CCBR Red Seminar Room, 160 College Street