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HST.161 Molecular Biology and Genetics in Modern Medicine  
Fall 2007

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There are two assigned papers.

I. Christof Stamma,\* , Ingeborg Friehsa, Siew Yen Hob, Adrian M. Moranc, Richard A. Jonasa, Pedro J. del Nidoa Eur J Cardiothorac Surg. 2001 Feb;19(2):195-202. Congenital supraaortic stenosis: a simple lesion?

II. Meyer-Lindenberg A, Mervis CB, Berman KF. Nat Rev Neurosci. 2006 7(5):380-93. Neural mechanisms in Williams syndrome: a unique window to genetic influences on cognition and behaviour.

Questions for reading assignment 3

1. What is the strongest evidence that the cause of SVAS in patients with William's syndrome is haploinsufficiency for the elastin gene and no other gene?

2. Explain how FISH could be used to obtain the data diagrammed in Figure 1c of the Meyer-Lindenberg et. al. paper. What reason could you suggest for the uncertainty in the extent of the deletion in most of the untypical cases presented in this figure.

3. What evidence supports the hypothesis that haploinsufficiency for the LIMK1 gene is responsible for the William's syndrome cognitive profile? What data is in conflict with this hypothesis?