

***Expression profile of FSHD supports a link between retinal vasculopathy and muscular dystrophy.*** Authors: Osborne RJ, Welle S, Venance SL, Thornton CA, Tawil R.

Paper published by Registry Co-investigators and colleagues at the University of Rochester.

Researchers continue to investigate the mechanisms of FSHD. The development of a more thorough understanding of the biological processes that are disrupted in FSHD has been a central goal for scientists involved in FSHD research for many years.

It has been suggested that the DNA deletion on chromosome 4 affects the functionality of the genes surrounding the deletion area. Chromosomes are “packages” in cells that contain genes and other DNA. Genes are “instructions” that the body uses to make proteins.

To explore this theory, researchers at the University of Rochester Medical Center collected tissue and blood samples from patients with FSHD as well as from patients with DM1 and subjects that were not affected with a muscle disease. They compared the activity of genes in skeletal muscle from the 3 groups of subjects. The researchers found that FSHD patients had an increase in gene expression (activity) in 44 different genes. Eleven of the genes have a role in the vascular (blood vessels) tissue of muscle and other cells. This increased activity in vascular genes was particularly notable in patients in the early stages of FSHD. These results may provide insight into the development of problems in the vascular tissue of the eye that commonly affect individuals with FSHD.

More information can be found at:

[http://www.ncbi.nlm.nih.gov/pubmed/17151338?ordinalpos=2&itool=EntrezSystem2.PEntrez.Pubmed.Pubmed\\_ResultsPanel.Pubmed\\_RVDocSum](http://www.ncbi.nlm.nih.gov/pubmed/17151338?ordinalpos=2&itool=EntrezSystem2.PEntrez.Pubmed.Pubmed_ResultsPanel.Pubmed_RVDocSum)