

## Multiple genes regulating the extracellular matrix are involved in groin hernia development *a systematic review of genetic and transcriptomic evidence*

**Stina Öberg<sup>1</sup>**, Emilie S. Engdal<sup>2</sup>, Ann H. Sæter<sup>1</sup>, Finn C. Nielsen<sup>3</sup>, Jacob Rosenberg<sup>1</sup>

1. Center for Perioperative Optimization, Department of Surgery, Herlev Hospital, University of Copenhagen, Denmark










2. Centre for Genomic Medicine, Rigshospitalet, Copenhagen, Denmark

3. Department of Clinical Medicine, University of Copenhagen, Denmark

### Aim

To evaluate the potential role of gene alterations in the development of groin hernias.


### Methods


-  Systematic review
-  Groin hernia versus control, any age
-  Biological samples to investigate genetic and transcriptomic data
-  Outcome: provide overview of
  -  gene expression (mRNA) and
  -  identify associated genes (DNA) that also theoretically could contribute to groin hernias
-  PubMed, Embase OVID, and CENTRAL
-  Bias: Quality of genetic association studies tool
-  Results were synthesised narratively and through *in silico* analyses


### Results

#### Case-control studies mRNA + DNA

20 case-control studies


 6 to 11,021 children and adults with and without inguinal hernia

 Inguinal hernia: possible alterations in genes regulating the closure of the processus vaginalis.


 Inguinal hernia: evidence of altered expression of procollagen 1 and 3 and enzymes involved in extracellular matrix homeostasis.


### GWAS

6 genome-wide association studies (GWAS)

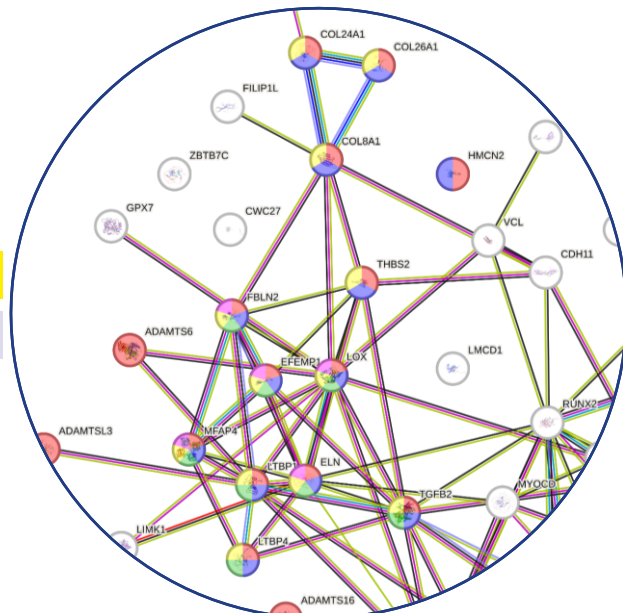
 75,805 to 552,102 adults with and without inguinal or femoral hernia

The GWAS identified genetic variants more common in patients with groin hernias than in controls and mapped these to genes.

 200 genes associated with inguinal hernia

 9 genes associated with femoral hernia

Our *in silico* analyses highlighted 33 genes associated with inguinal hernia, mainly involved in extracellular matrix organisation, including collagen and elastin formation.



*STRING network based on the GWAS data: interactions between proteins encoded by genes associated with inguinal hernia. Coloured genes are involved in extracellular matrix formation.*

### Conclusion

More genetic and transcriptomic data are needed in children.