Collagen alpha-1(XVIII) chain is a protein that in humans is encoded by the COL18A1 gene

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Caglayan et al. investigated the protein expression pattern of COL18A1 in the mid-fetal and adult human cerebral cortex and then analyzed the spatial and temporal changes in the expression pattern of COL18A1 during human cortical development using the Human Brain Transcriptome database.

They identified two novel homozygous deleterious frame-shift mutations in the COL18A1 gene. On further investigation of these patients and their families, they found that many exhibited certain characteristics of Knobloch syndrome, including pronounced ocular defects. This data strongly support an important role for COL18A1 in brain development, and this report contributes to an enhanced characterization of the brain malformations that can result from deficiencies of collagen XVIII.

This case series highlights the diagnostic power and clinical utility of whole-exome sequencing technology-allowing clinicians and physician scientists to better understand the pathophysiology and presentations of rare diseases. They suggest that patients who are clinically diagnosed with Knobloch syndrome and/or found to have COL18A1 mutations via genetic screening should be investigated for potential structural brain abnormalities even in the absence of an encephalocele [1)].

1)
Caglayan AO, Baranoski JF, Aktar F, Han W, Tuysuz B, Guzel A, Guclu B, Kaymakcalan H, Aktekin B, Akgumus GT, Murray PB, Erson-Omay EZ, Caglar C, Bakircioglu M, Sakalar YB, Guzel E, Demir N, Tuncer O, Senturk S, Ekici B, Minja FJ, Šestan N, Yasuno K, Bilguvar K, Caksen H, Gunel M. Brain malformations associated with Knobloch syndrome–review of literature, expanding clinical spectrum, and identification of novel mutations. Pediatr Neurol. 2014 Dec;51(6):806-813.e8. doi: 10.1016/j.pediatrneurol.2014.08.025. Epub 2014 Sep 4. PMID: 25456301; PMCID: PMC5056964.