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Important discovery for the diagnosis of genetic diseases

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A study conducted by Marie Kmita's team at the IRCM, in collaboration with Josée Dostie at McGill University, shows the importance of the chromatin architecture in controlling the activity of genes, especially those required for proper embryonic development. This discovery, recently published in the scientific journal *PLOS Genetics*, could have a significant impact on the diagnosis of genetic diseases.

Each cell in the body contains a person's genetic information in the form of DNA molecules, wrapped around structures called nucleosomes. Together, the DNA and nucleosomes form the chromatin, which is the main component of chromosomes.

"Our work shows that the regulation of the activity of genes controlling embryonic development is linked to the three-dimensional organization of the chromatin," explains Dr. Kmita, Director of the Genetics and Development research unit at the IRCM. "In fact, this chromatin architecture, which varies according to the cell type, generates specific contacts between sequences of regulatory DNA and the genes they regulate."

To date, studying the causes of genetic diseases is mainly achieved through DNA sequencing and the analysis of gene sequences. However, the cause of such diseases could just as well be an anomaly in the DNA sequences that control the genes.

"It is now possible to identify regulatory DNA that controls a given

gene," adds Dr. Kmita. "Our discovery paves the way for studying the mechanisms that control the architecture of chromatin, which should have a significant impact on identifying the causes and diagnosing genetic diseases."

The IRCM researchers' scientific breakthrough could have an impact on a large number of genetic diseases, including those associated with the Hox genes studied by Dr. Kmita, such as synpolydactyly (a congenital malformation characterized by the fusion of digits and the production of additional digits) and the hand-foot-genital syndrome (a genetic disease characterized by limb malformations and urogenital defects).

More information: www.plosgenetics.org/article/i … journal.pgen.1004018

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