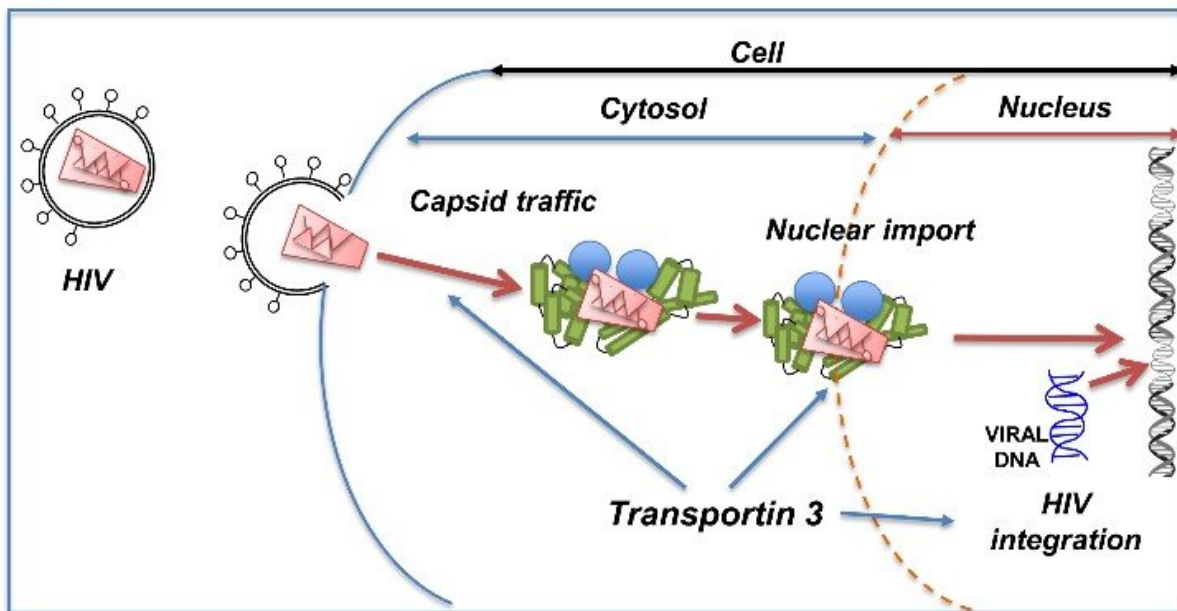


Researchers identify second gene mutation linked to HIV resistance

August 29 2019

ROLE OF TNPO3 IN HIV INFECTION



Role of TNPO3 in HIV Infection. Credit: Rodríguez-Mora S, et al. (2019)

A rare genetic mutation that causes a form of muscular dystrophy affecting the limbs also protects against HIV infection, Spanish scientists reported Thursday.

The breakthrough comes a decade after American Timothy Brown,

known as the "Berlin Patient," became the first person cured of HIV after a bone marrow transplant from a donor with a mutation of the CCR5 gene.

The newly-discovered mutation concerns the Transportin 3 gene (TNPO3) and is far more rare.

It was identified several years ago among members of a family in Spain who were suffering from type 1F limb-girdle muscular dystrophy.

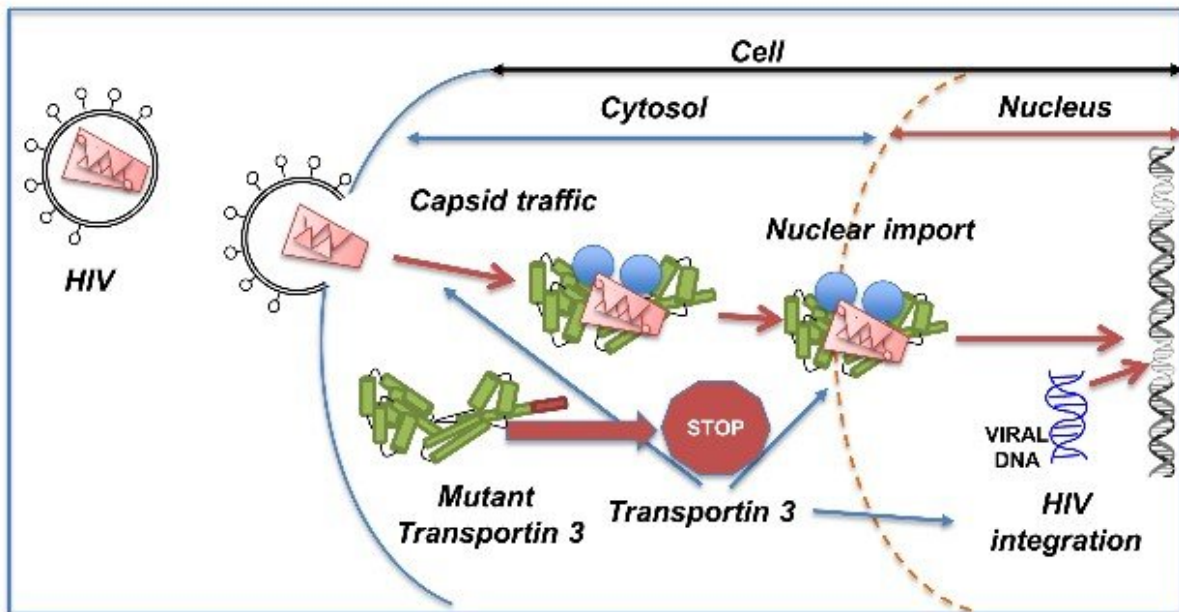
Doctors studying the family learned that HIV researchers were interested in the same gene because it plays a role in transporting the virus inside cells.

They then got in touch with geneticists in Madrid, who took blood samples from those family members and infected the blood with HIV—revealing a welcome surprise.

The lymphocytes—white blood cells that are an important part of the immune system—of people with the rare muscular illness were naturally resistant to HIV, it emerged.

"This helps us to understand much better the transport of the virus in the cell," Jose Alcamí, a virologist at the Carlos III Health Institute and co-author of a paper published in US journal *PLOS Pathogens* on the subject, told AFP.

ROLE OF TNPO3 IN HIV INFECTION AND BLOCK BY MUTATED TNPO3



Role of TNPO3 in HIV Infection and block by mutated TNPO3. Credit: Rodríguez-Mora S, et al. (2019)

HIV is among the most studied viruses, he said, adding however that much remained to be learned, such as why five percent of patients who are infected do not develop AIDS.

"There are mechanisms of resistance to infection that are very poorly understood," he said.

More information: Rodríguez-Mora S, De Wit F, García-Perez J, Bermejo M, López-Huertas MR, Mateos E, et al. (2019) The mutation of Transportin 3 gene that causes limb girdle muscular dystrophy 1F induces protection against HIV-1 infection. *PLoS Pathog* 15(8): e1007958. doi.org/10.1371/journal.ppat.1007958

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Citation: Researchers identify second gene mutation linked to HIV resistance (2019, August 29)
retrieved 27 April 2024 from

<https://medicalxpress.com/news/2019-08-mutation-rare-muscle-disease-hiv-.html>

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