Diciembre 2020

Boletín bibliográfico

Enfermedad de Machado-Joseph

La ataxia es una enfermedad rara de origen genético que se caracteriza por alteraciones neuromusculares debidas a la pérdida selectiva de neuronas en el cerebelo, el órgano de nuestro sistema nervioso encargado de controlar el movimiento y el equilibrio.

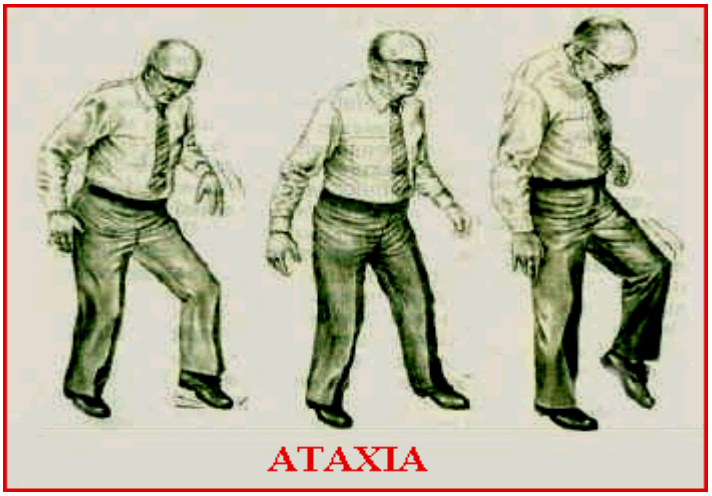
Investigadores de la Universidad de Barcelona (UB) han identificado nuevas funciones del gen ataxina 3 (ATXN3) --causante de la enfermedad de Machado-Joseph, el tipo de ataxia más frecuente-- en el desarrollo de los fotorreceptores de la retina.

Según los investigadores, estos resultados son relevantes no solo para profundizar en las causas moleculares de la ataxia y en el diseño de potenciales terapias contra ella, sino también para comprender otras enfermedades, como las degeneraciones maculares.

La enfermedad de Machado-Joseph, también llamada ataxia espinocerebelosa de tipo 3, está causada por mutaciones dominantes de ganancia de función del gen ATXN3.

Dichas mutaciones inducen la formación de agregados neurotóxicos que conllevan la muerte progresiva de las neuronas cerebelares. No obstante, se sabe muy poco de cuál es la función básica del gen ATXN3.

Amazings



SCA 3

- SCA3 o Enfermedad de Machado Joseph (EMJ) es la SCA mas frecuente en la mayoría de poblaciones.
- Se ha descrito como ataxia cerebelosa pura, parkinsonismo familiar, paraplegia espástica hereditaria, neuropatía hereditaria y síndrome de piernas inquietas.
- Tiene una frecuencia relativa del 1-63% y se debe a la mutación en el locus 14q32.1, con una expansión de 51-89 tripletes CAG

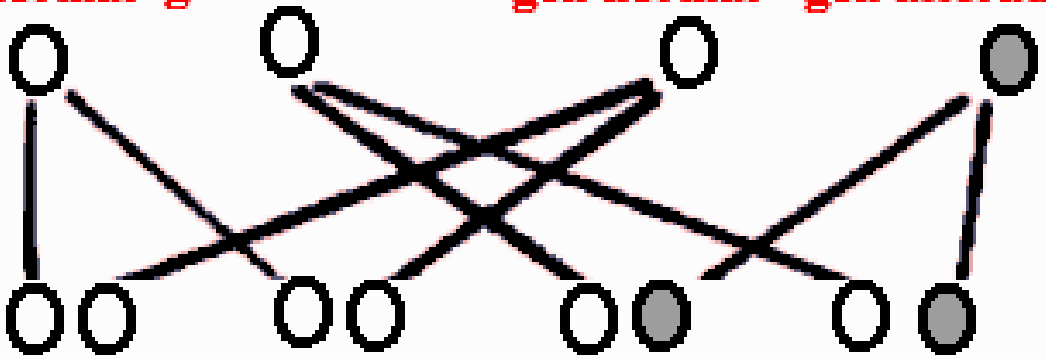
TRANSMISIÓN DOMINANTES (teorías)

<=====>

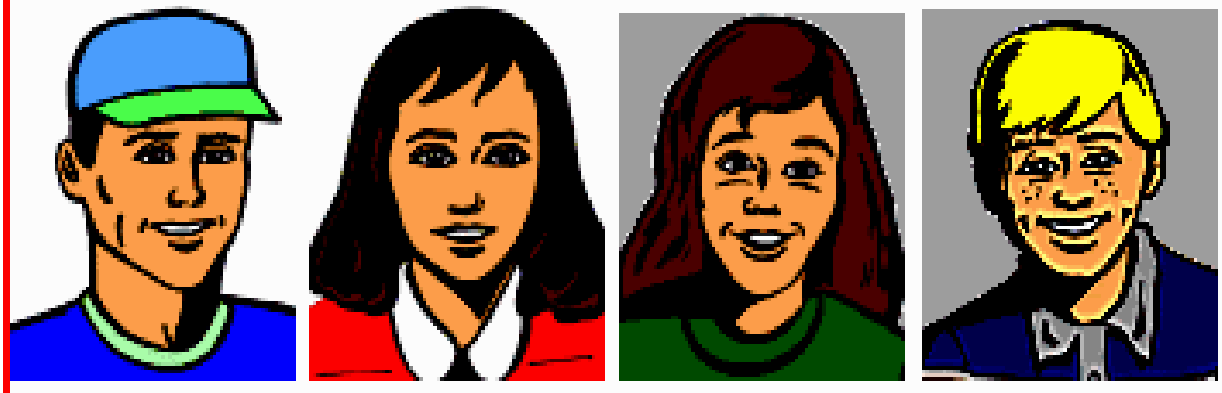
Un solo gen defectuoso basta para ser afectado

NORMAL — **AFECTADA**

gen normal gen normal gen normal gen alterado



NORMAL NORMAL AFECTADA AFECTADO



25 % + 25 = 50 %

25 % + 25 % = 50 %

La incidencia por sexos, el orden, y los porcentajes reales, quedan sujetos al azar y sin posibilidad de ser definidos

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