Should We Call Them The Living Dead? هل يجب تسميتهم الموتي الاحياء؟

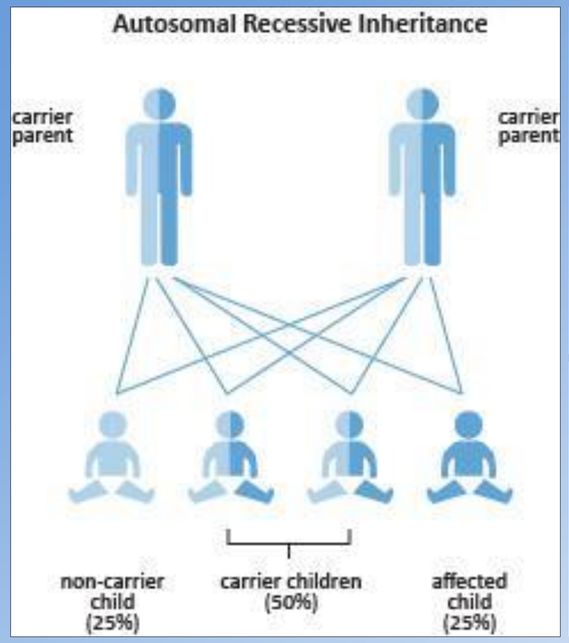
Libyan International Medical University

Faculty of Basic Medical Sciences

By: Hamdy Jawdat Alashker /2nd Year Medical University

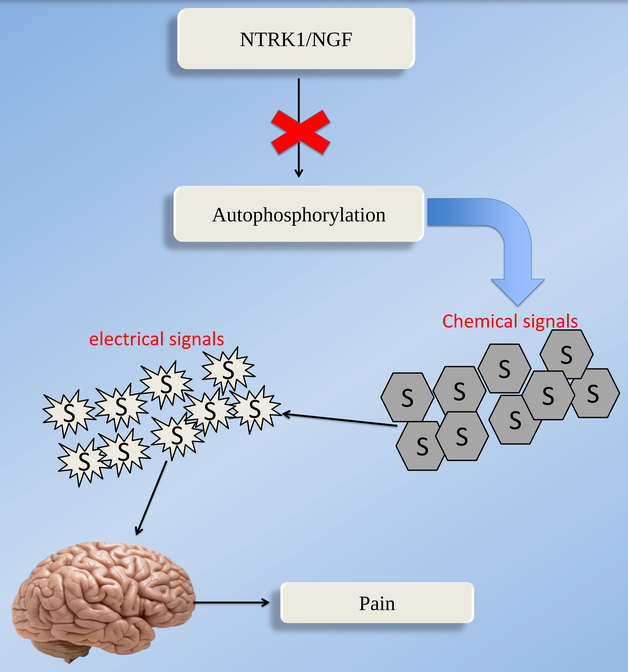
What's Congenital Insensitivity to Pain (CIPA)?

Congenital insensitivity to pain (CIPA), also called hereditary sensory and autonomic neuropathy (HSAN), is a rare neurological disorder of the nervous system with no cure.
It is an autosomal recessive disorder - a genetic mutation in an autosome (a non-sex chromosome) that only occurs when either both parents are affected or are carriers of the recessive genes which are then passed on homozygously to the child.
It is commonly accompanied by hyperactivity and anhidrosis. CIPA results in the patient's inability to sense extreme coldness, heat, or pain and even nerve related sensations such as hunger and the need to urinate . The lack of pain patients experience often leads to self-mutilation, bone fractures and eye Damage.[1]



Cause of CIPA:-

The cause of CIPA has been tracked to mutations in the **NTRK1/NGF** receptor gene, including the G571R and the R774P. Functioning normally, this receptor, upon receiving the nerve growth factor, starts a chain of autophosphorylation, which then sends a signal to the brain. However, in a patient with CIPA, the aforementioned gene is mutated in a way that interferes and halts the autophosphoylation process, therefore stopping signals of pain and temperature from being sent to the brain.[2]



Statistics:-

- CIPA is so hard to study because the majority of patients die before age 3 because of fever Since it was first reported in 1983, there have been less than 60 documented cases in the United States. Currently there are 17 living people diagnosed in the US.

- There are, however, over 300 documented cases in Japan. In the world population, CIPA is most common among Ashkenazi Jews. The reason for both of these is because it is more likely to occur in homogenous societies.[3]

Living with CIPA

Patients with CIPA can live a fairly normal life. However, there are certain precautions that need to be taken in order to ensure the safety of the patient. The patient must constantly check for cuts, bruises, self-mutilations, and other possible unfelt injuries. Also, they must regularly visit their doctor to receive a full work up to make certain that they do not have any internal problems that can be fatal to them (Insensitive). also a wheelchair might be necessary as a result of deteriorating joints.

Another precaution that must be taken by the patient is checking their temperature. The fact that they are unable to sweat means that they have a higher risk of fever because they are unable to cool themselves down. This fever can often lead to mental retardation and death. They must drink a lot of water, to make sure that they are not dehydrated.[1]

Conclusion:-

The imagination of unable to feel pain can consider one of the worst problem in the world. Pain is our limit that tell us there is something wrong and we need to stop even though if the pain was physical or emotional.

References:-

1- Katie Lambert "How CIPA Works" 21 September 2007. HowStuffWorks.com. <http://science.howstuffworks.com/life/inside-the-mind/human-brain/cipa.htm> 2 April 2017

2- Indo Y. Molecular basis of congenital insensitivity to pain with anhidrosis (CIPA): mutations and polymorphisms in TRKA (NTRK1) gene encoding the receptor tyrosine kinase for nerve growth factor. Hum Mutat.2001 Dec;18.

3-Verhoeven K, Timmerman V, Mauko B, Pieber TR, De Jonghe P, Auer-Grumbach M. Recent advances in hereditary sensory and autonomic neuropathies. Curr Opin Neurol. 2006 Oct;19

نبذة عن الموضوع:-

- سيبا هو خلل عصبي وراثي يتميز حامله بفقدان الاحساس بالالم والتعرق.

- الخلل نتيجة الي تشوه في المستقبلات المسؤولة عن نقل احساس الالم الي الدماغ نتيجة لذلك لا يصبح قادر علي تفسير الالم.

- احصائيا تم توثيق 60 حالة في الولايات المتحدة و 300 حالة في اليابان.

- المريض لا بد ان يتخذ احتياطات وقائية للحفاظ علي سلامته مثل فحص شامل لجسمه يوميا بحثا عن كدمات او جروح وزيارة طبيبه بإنتظام.