PHARC syndrome, a rare inborn error of phospholipid metabolism disease is characterised by Progressive Hearing loss, , neuropathy and Ataxia, Retinitis pigmentosa and early onset Cataract.

However, patients do not always present all of these symptoms at diagnosis. This pathology is the result of homozygous or heterozygous composite mutations of ABHD12. Here we report the story of a 45-year-old patient with hearing loss associated with retinitis pigmentosa and bilateral cataract due to a homozygous mutation of ABHD12. This patient is also a high level athlete and has neither ataxia nor polyneuropathy.

**Patient phenotype**

45 years old male, Portuguese parents not known to be related

Sporadic case, one healthy brother

Polyneuropathy: none

Hearing Loss: moderate bilateral progressive hearing loss, diagnosed at 12, bilateral hearing aids at 31. Tinnitus +++ . No dizziness

Ataxia: none

Retinitis Pigmentosa: diagnosed at 17, stop driving at age 35

Cataract: bilateral

Homozygous non-sens mutation of ABHD12 (p.Arg352*)

**A prolific sporting career**

The patient started the road cycling competition at 21 years. He performed more than 15 cycling races of more than 200km.

Longest distance covered: Paris-Faro race 2300km.

Running: started at the age of 43 (Millau ultramarathon 100km)

Triathlon: started at the age of 44.

The sports addiction screening questionnaire shows a strong appetite for effort without addiction.

**Discussion**

This gene codes for ABHD12 a protein involved in the endocannabinoid pathway that is central to addiction mechanisms. Furthermore, an accumulation of proinflammatory lipids has been demonstrated in the mouse model of PHARC syndrome. This accumulation is held responsible for a large part of the symptoms. The p.Arg352* mutation has already been reported in one patient with ataxia and early polyneuropathy. The absence of neurological impairment in our patient suggests the presence of modifying environmental factors. One of these factors could be the intense sporting activity that helps prevent neuroinflammation. Moreover, the deregulation of endocannabinoid metabolism could be a factor promoting intensive sports practice.

**ABHD12: transmembrane protein**

2 known functions

- hydrolyzation of 2-arachidonyleglycerol
- lysophosphatidylserine lipase activity.

Mouse and zebrafish models present with similar phenotypes.