

## Neurofibromatosis type 1 (Von Recklinghausen's disease) in Emergency Department

Domingo Ly-Pen<sup>\*1</sup>, Diana Ly Liu<sup>2</sup>

<sup>\*1</sup>Emergency Department, Hospital Universitario Ramón y Cajal, Carretera Colmenar Viejo, Km 9, 28031, Madrid, Spain, Croydon UCC, Croydon University Hospital, 530 London Road, Croydon CR7 7YE, UK

*Email id - domingoly@gmail.com*

<sup>2</sup>Anaesthesiology Department, Hospital Universitario Basurto, Montevideo Etorbidea, 18, 48013 Bilbao, Vizcaya, Spain

*Email id - dianalyliu@gmail.com*

### Abstract:

*Neurofibromatosis is one of the most common genetic disorders. A typical picture is presented, with a brief compilation of the most common presentations of these patients in the Emergency Department.*

*Keywords: Neurofibromatosis type 1, Von Recklinghausen's disease, Emergency Department.*

### TEXT:

Neurofibromatosis is one of the most common genetic disorders, autosomal dominant trait with a frequency of about 1 in 3000; it is inexorably progressive, but with markedly variable expressivity<sup>1</sup>.

Classified into two genetically different subtypes characterized by multiple cutaneous lesions (Figure 1) and tumors of the peripheral and central nervous system.



Neurofibromatosis type 1 (Von Recklinghausen's disease) is a tumor disorder due to a mutation of a gene on chromosome 1.

Peripheral nervous system is usually the primary neurologic involvement in Von Recklinghausen's disease. Central nervous system is secondarily involved.

In Emergency Department the most usual attendances are local infection around the fibromas.

Other reported causes in Emergency Department are: spontaneous hemothorax<sup>2</sup>, digestive hemorrhages due to gastrointestinal stromal tumors<sup>3</sup>, upper airway obstruction (due to neurofibroma in base of tongue<sup>4</sup>, ruptured subclavian artery aneurysm<sup>5</sup>).

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