

## **PS2.181**

### **Doctor I have a lump in my neck**

*Raquel Navarro Hernández(1), F Amorós Martínez(2), AM Magdaleno Hernández(1)*

*(1) Albaterra Health Center, Vega Baja Hospital, Orihuela, Spain*

*(2) Vinalopó Hospital, Elche, Spain*

*Corresponding author: Dr Raquel Navarro, Albaterra Health Center, Orihuela, Albaterra, Spain. E-mail: ranaherna@gmail.com*

**Background:** Female 39 year old with no medical history who complains of recurrent tonsillitis. On examination diffusely increased thyroid gland is palpated. Cervical TC was made with the result of multinodular goitre. The PAAF showed follicular proliferation. Total thyroidectomy was performed and the anatomic pathological result was multinodular hyperplasia and multiple adenomatous nodules that recommended discard Cowden Syndrome although PTEN was negative.

**Method:** S Genetic study confirmed Cowden Syndrome. Study was completed by dermatological exploration, mammography, endoscopy and colonoscopy, chest-abdominal TC, uterine ultrasound, vaginal and urine cytology, and encephalic MNR.

**Results:** The skin inspection showed multiple lentigos, seborrheic keratosis, and various trichilemmomas. The colonoscopy result was four polyps, three of them were hyperplastic polyps and only one was an hamartoma. All the other diagnosis test resulted normal.

**Conclusions:** Cowden disease is an autosomal dominant condition with variable expression that can be associated with a mutation in the PTEN gene on arm 10q, that causes hamartomatous neoplasms of the skin and mucosa, GI tract, bones, CNS, eyes, and genitourinary tract. Skin is involved in 90-100% of cases, and the thyroid is involved in 66% of cases. The prevalence is estimated to be approximately 1 case per 200,000 population; however, it is likely more prevalent because many features of Cowden disease are found in the general population and the diagnosis may be overlooked, which leads to underdiagnosis. Penetrance is thought to be nearly complete; it approaches 90% by age 20 years. The importance of the genetic advice and early diagnosis resides in its primary association with increased frequency of malignant tumors, 25-50% breast cancer, 3-10% thyroid cancer and 5-10% endometrial cancer principally.