Radiocubital Synostosis: Malformation Behind Klinefelter Syndrome

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Radiocubital synostosis (RCS) – figure 1, is a rare malformation of the upper limb, with fusion of the radio and ulna.

There are two forms: acquired or post-traumatic and congenital.

The last form can be isolated or secondary to other malformations or syndromes.

Klinefelter syndrome is the group of chromosomes disease in which at least one of the X chromosome is add to the male karyotype.

Is the most common aneuploid chromosome with a incidence of 1:650 in male individuals.

The most common symptoms are: small testis, infertility, azoospermia, rare pubic and facial hair, learning disabilities, gynecomastia, language delay and tall stature.

The clinical manifestation can be subtle in early childhood and many times the syndrome is under-diagnosed.

RCS can be associated in individuals with Klinefelter syndrome, but when is happened appears in patients with karyotype 48, XXY and 49, XXXY, and rarely 47, XXY.

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Whenever we find a patient with isolated RCS, we cannot exclude that behind this malformation, we have a patient with Klinefelter. A simple karyotype can confirm this diagnosis.