Autoimmune thyroiditis in Klippel-Feil syndrome with Arnold Chiari and syringomyelia

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Introduction

Klippel-Feil Syndrome (KFS), prevalence of 1:40000, is characterized by congenital fusion of two or more cervical vertebrae; three major features are short neck, low hairline at the back of the head and a limited range of motion in the neck. The phenotypic expression is variable, presenting with other vertebral abnormalities (congenital high scapula, scoliosis, kyphosis, spina bifida, malformation of the atlas, hemivertebrae) or extra skeletal symptoms such us deafness, renal, heart or neurological abnormalities like neuroschisis and syringomyelia. Most cases are sporadic, but an autosomal dominant form, linked to a mutation in the gene GDF6 (8q22) or to a chromosomal rearrangement involving the long arm of chromosome 8, has been described.

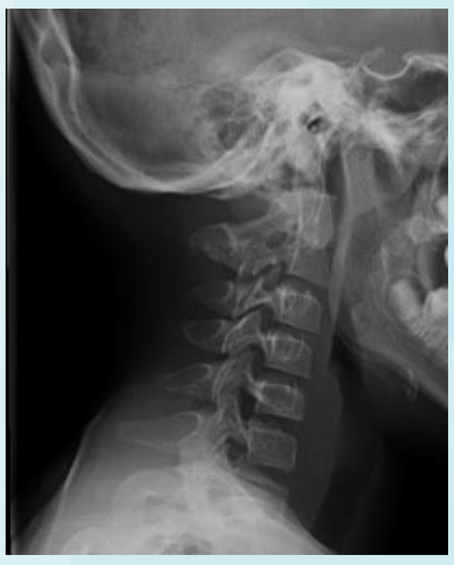
Case study

A girl was referred to our department at the chronological age (CA) of 10.3 yrs with a suspect of Turner syndrome, due to the presence of webbed neck and progressive deceleration of growth velocity. The girl, born in Russia, was adopted at the CA of 14 months. Clinical examination showed short neck, thelarche Tanner stage II, pubarche Tanner stage II, hyperlordosis and valgus elbow. The height was 134 cm (-1.1 SDS), weight 28 kg (-1.3 SDS).

Pelvic ultrasound showed normal uterus and ovaries, karyotype was not performed.

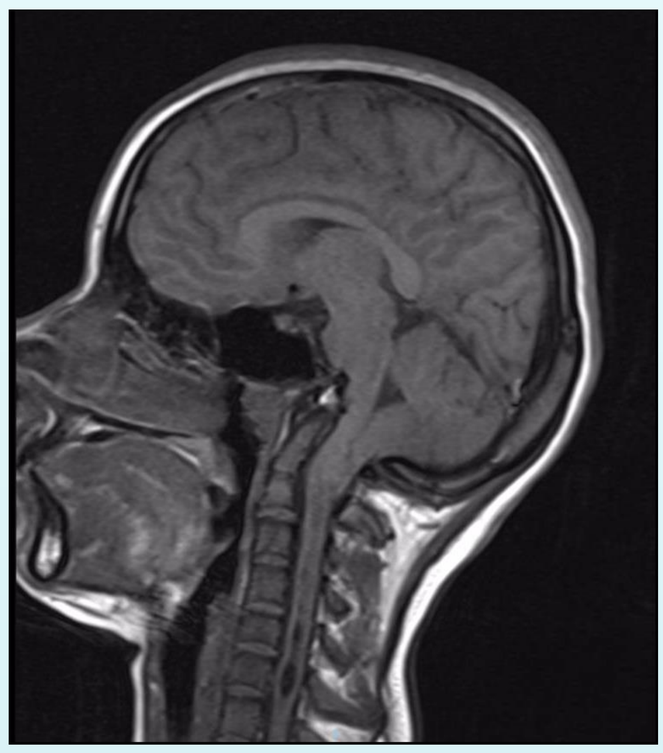
Due to the presence of short neck, cervical x-ray was performed: the images showed fusion of the cervical vertebrae C2 and C3, confirming the suspect of a Klippel-Feil syndrome. Figure 1

Figure 1



The brain MRI showed Arnold-Chiari (AC) type I malformation associated to syringomyelia from C2 to C7. Figure 2

Figure 2



Blood investigations showed markedly elevated TSH values(> 75 µU/ml, range 0.4 -4.0) and low valuesof FT3 (2.5 pg/ml, range 2.5-3.9) and FT4 (4.1 pg /ml, range 5.8-16.4).

Serum antitireoglobulin and antiperoxidase antibodies were both elevated: ATG > 3000 U/ml (range < 45), ATPO 298 U/ml (range <35).

Thyroid ultrasound showed increased thyroid size with markedly heterogeneous and hypervascular echogenicity with slightly thickened isthmus. The patient was started on thyroid replacement therapy.

Conclusion

To our knowledge, association of KFS with AC malformation, syringomyelia and autoimmune thyroiditis has not been reported in the literature. We believe that the presence of thyroiditis in our patient represents a random association, since neither in our patient nor in the literature there are data supporting the hypothesis of an increased incidence of autoimmune disorders in these patients. Nevertheless, we suggest to evaluate also thyroid function in these patients to rule out the presence of abnormalities.

References

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