ORIGINAL RESEARCH PAPER

Radio-Diagnosis

ROLE OF USG AND MRI IN DE LA CHAPELLE SYNDROME

KEY WORDS: Male phenotype, small testes, 46 XX, SRY gene

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ABSTRACT

De La Chapelle syndrome is a rare condition where individuals with male phenotype have 46 XX chromosome in all cells. They usually present in adolescent or adult age group with chief complaints of small size of testis and/or penis or infertility with gynecomastia in one third cases. Imaging modalities like USG and MRI help confirm the absence of Mullerian structures and the presence of seminal vesicle and prostate, followed by which chromosome studies showing 46 XX chromosome confirms the diagnosis.

INTRODUCTION

De la Chapelle syndrome, also known as 46 XX male, is a rare disorder seen in 1:20000 new born males.(1) Testis Determining Factor (TDF), located on the short arm of the Y chromosome, is responsible for testicular development in males. Sertoli cells secrete Müllerian Inhibiting Factor which is responsible for the agenesis of Müllerian structures. That action, along with Leydig cells that secrete testosterone, ultimately lead to male internal genitalia development. (1) In the majority of the cases of this syndrome, Y chromosome is translocated on X chromosome as the result of recombination in the distal parts of short arms of X and Y chromosomes during paternal meiosis (SRY-positive). (1).

Other explanations are - (i) X-linked mutation/ overex pression in the genes that cause testis differentiation or mutation/overexpression in autosomal genes [e.g. SRY box-related gene 9 (SOX9)] in SRY negative XX males, and (ii) secret Y mosaicism found only in the gonads (2) Three groups have traditionally been described, based on phenotype: males with normal male phenotype, males with ambiguous genitalia, and true hermaphrodites. Male phenotype, small testes, and azoospermia are found in most cases. Gynecomastia may be associated with one-third of all patients, while low height, cryptorchism, and hypospadias are less frequently seen (3). Most patients present with complain of small testis and penis or with infertility. Testosterone supplementation at an early age is the primary treatment for this condition. Early imaging in patients with small testes can help towards reaching the diagnosis and hence early treatment.

Case Study

In the present case, a 17 year old male presented with complaints of small size of testis and penis. Patient had a normal male phenotype and no complaints of gynecomastia. Semen analysis showed azoospermia.

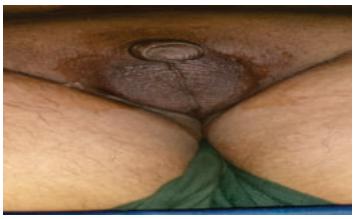

Figure 1- hypoplastic testes in scrotal sac and small penis

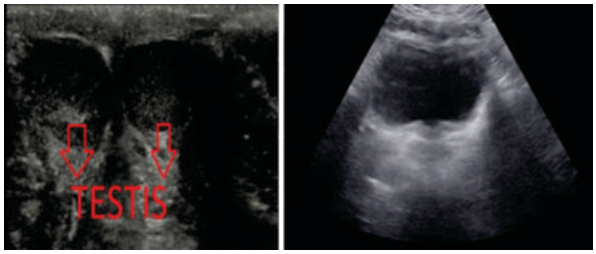

Figure 2-
USG showed normal prostate and small testes like structure in scrotal sac.Uterus and ovaries were absent.

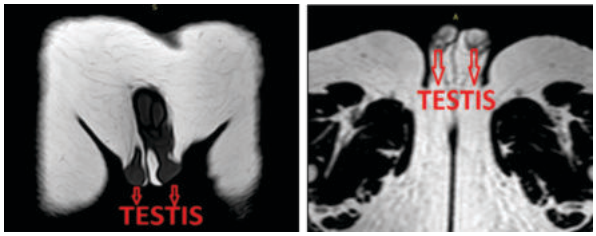

Figure 3-
MRI of abdomen-pelvis showed two testes like ovoid structures in hypoplastic scrotum on both sides, measuring 11x20x20 mm on right side and 12x21x21 mm on left side suggestive of hypoplastic testes. Normal prostate and seminal vesicle were noted.

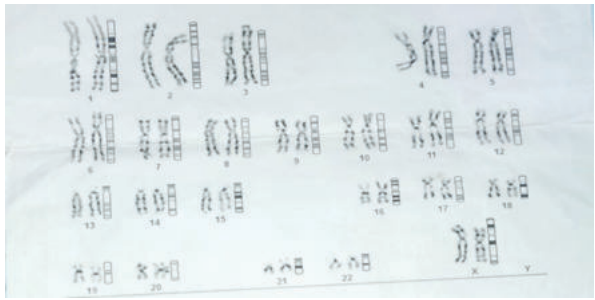

Figure 4 Karyotype revealed 46 XX chromosome.

CONCLUSION

Final diagnosis is made by combination of clinical features, imaging and karyotyping- phenotypically male individual with absent uterus and ovaries and presence of prostate and 46 XX chromosome - de la Chapelle syndrome.

Early imaging in patients with these complaints can help confirm the absence of Mullerian structures, therefore raising a high suspicion of this syndrome.

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