

# COEXISTENCE OF OVARY TUMOR, CONGENITAL ADRENAL HYPERPLASIA AND TRIPLE TRANSLOCATION INVOLVING CHROMOSOME 9,11 AND 12: INITIAL STEPS OF DEFINING NEW SYNDROME?

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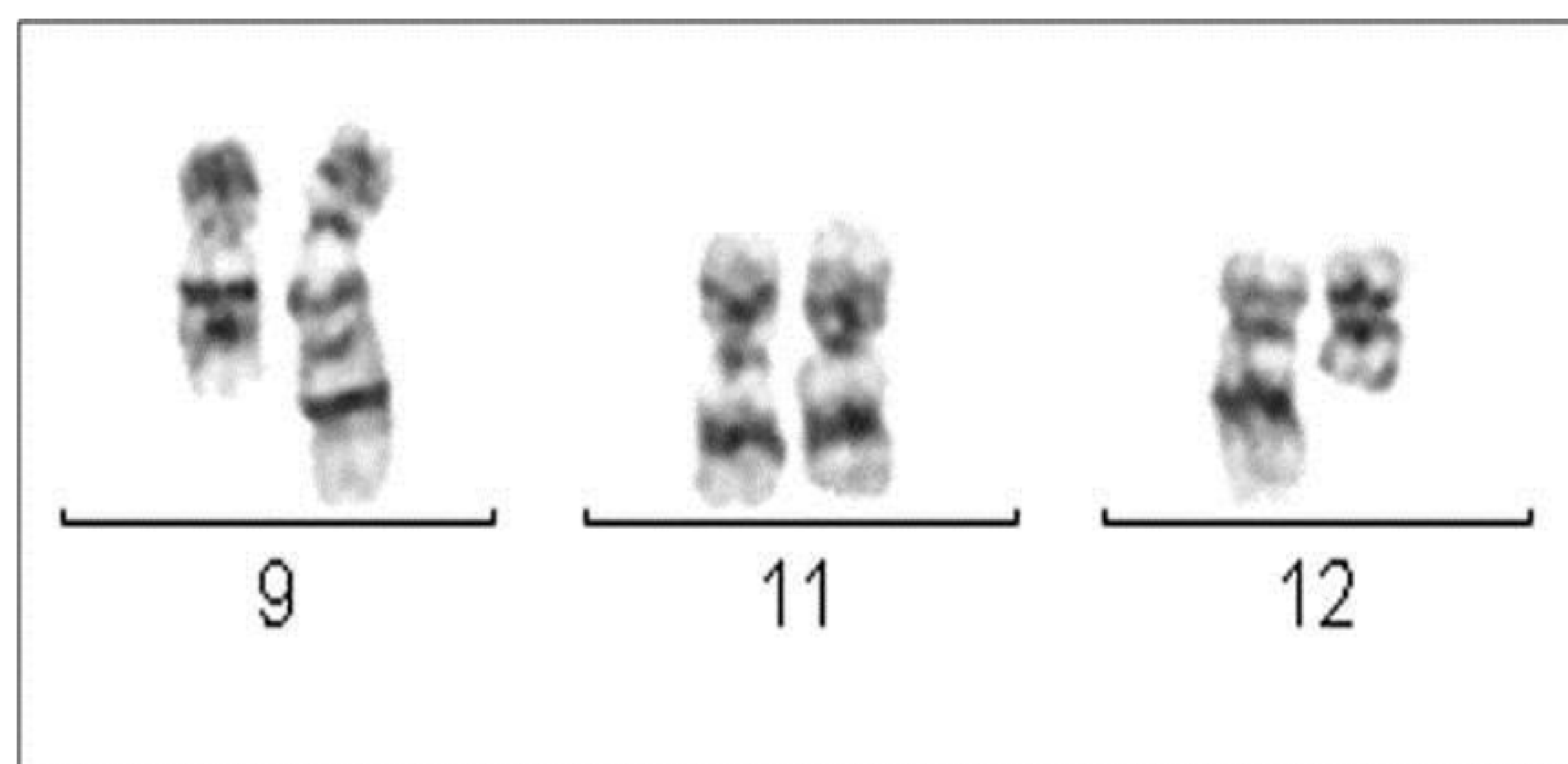
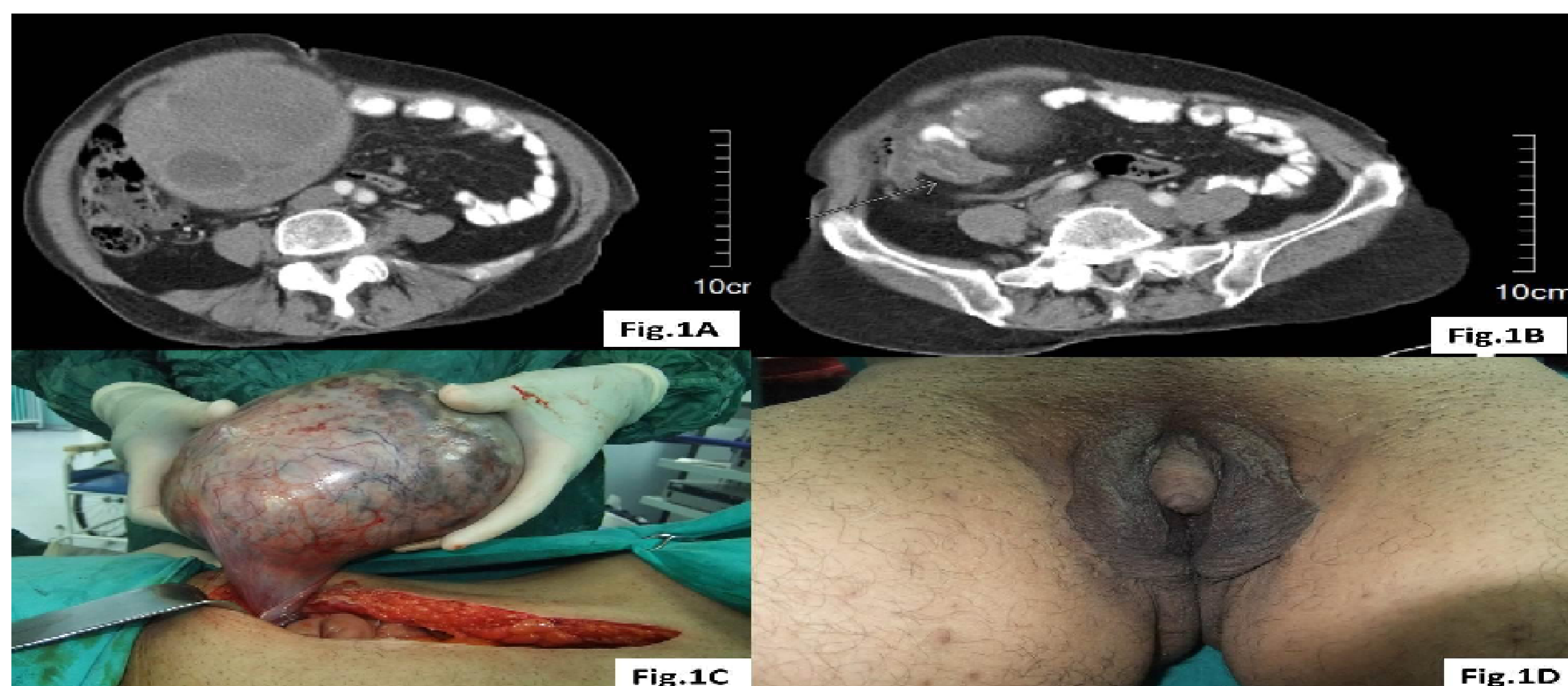
## INTRODUCTION AND AIM:

Congenital adrenal hyperplasia (CAH) is rare autosomal recessive disease. CAH due to 21-hydroxylase deficiency accounts for 95% of cases. We aim to define in this report 59-year-old woman with simple virilizing type CAH who diagnosed granulosa cell tumor and I172N mutation in the CYP21A as well as triple translocation involving chromosome 9p,11p and 12p at first time in the literature.

## CASE REPORT:

A 59-year-old woman was applied to our clinic complained with abdominal pain and distension. She had irregular menstrual cycle per 3 to 4 month and hypomenorrhea. She had gone through the menopause at 40 years old. She had never become pregnant. In physical examination her breast tissue had not developed and virilism was determined (modified Ferriman Galvey score >16). A palpable solid mass was detected in the right of abdomen. Ambiguous genitalia, clitoramegaly and hyperpigmentation were found. In computerized tomography giant ovarian mass in mesentery tissue was detected. The mass (3500 gram) was removed with the right tubo-ovarian structures. The granulosa cell tumor was diagnosed by the immunohistochemical examination.

The high serum concentration of 17-OH progesteron measured at baseline and after 250- $\mu$ g bolus of synthetic ACTH. In genetic analysis, we screened for six point mutations, large deletions, and noncommon mutations using restriction fragment length polymorphism (RFLP) methods, PCR, and sequencing of CYP21 gene respectively. The patient was detected to be homozygote for the I172N mutation. In addition, fifty percent of the metaphases examined had triple translocation [t(9;11;12)] between chromosomes 9p, 11p and 12p.



**CONCLUSION:** The I172N mutation in the CYP21A accompanied with t(9;11;12) translocation did not define previously in patients with CAH. This mutation may be sign of a new syndrome or a co-insidans that triggered to granulose cell tumor.

