them had normal adrenal function. Furthermore, testicular function was evaluated in all patients. the karyotype was 46,XY for all patients. The syndromic one had additional mutation in FRAS1 gene besides NR5A1 mutation which might cause the associated renal manifestations. additionally, the last two patients had the same NR5A1 novel mutation.

Conclusion: Although, NR5A1 gene mutation is a common cause of XY DSD, it has a wide spectrum of phenotypic features. This may be due to different mutations in the same gene or due to associated gene mutations.

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Oligomenorrhea in two girls with familial mediterranean fever: how chronic inflammation can impair ovarian cycle

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Introduction: Familial Mediterranean Fever (FMF) is an inherited auto-inflammatory disorder still extremely underdiagnosed in the Mediterranean area.

The disease is secondary to a gain of function mutation of the MEFV gene, classically defined as "autosomal recessive", with possible symptoms also in heterozygous patients. The mutation induces a hyperexpression of IL-1 beta and a chronic inflammation. Clinical manifestations are characterized by recurrent attacks of fever, polyserositis, abdominal, thoracic, articular pain, fatigue, rash, oral aphthae, myalgia, etc. Some young girls suffer of oligomenorrhea or amenorrhea, especially in cases of unsatisfactory control of chronic inflammation.

Treatment is based on colchicine and, in non-responders, anti-IL-1 biological drugs.

Materials and Methods: We describe the case of two sisters, 16 and 18 years-old, affected by FMF (hererozygous MEFV gene mutation: A744S), with a limited number of attacks characterized by fever, abdominal pain, oral aphthae. Both showed a persistent increase of serum amyloid A (SAA) (> 13 mg/l) in the attacks-free intervals. Treatment with colchicine was followed by a rapid SAA level normalization. Following the well-being status, the patients decided to stop colchicine treatment. In the following years, both showed a persistent increase of SAA levels, with oligomenorrhea and anovulatory menses, with increased LH/FSH ratio.

They started again the treatment with colchicine (1 mg/day), with the reduction and normalization of SAA levels and the recovery of menses cycles within 6 months.

Conclusions: FMF can interfere with the gonadic function, compromising hypothalamic-hypophyseal-gonadic axis. The pathogenesis is linked to chronic subclinical inflammation and treatment with colchicine can normalize SAA, with the improvement of gonadic function. We highlight the importance of endocrine follow-up in patients with autoinflammatory syndromes, as FMF, to prevent gonadic impairment and infertility.

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Classical CAH girls having early intervention and puberty development

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Introduction: CAH (congenital adrenal hyperplasia) is the most common cause of ambiguous genitalia among girls. 21 Hydroxylase deficiency is the most common type of CAH. If the CAH girls have early intervention and they have started early treatment then they will achieve normal puberty. IF CAH girls have started treatment later in the life then they will present either with precocious puberty or delayed puberty.

Methodology: We have collected data from 27 girls who are diagnosed as classical CAH in our Endo OPD. Inclusion Criteria includes All girls who have ambiguous genitalia and diagnosed as 21-hydroxylase deficiency and are 46XX, these girls are 15 years or older. Exclusion Criteria Includes non classical CAH, CAH girls reared as boys,46XY. Detail history, physical examination, lab investigation and medication record were noted and verified from the parents as well.A questionnaire was made which includes age of presentation in endo clinic, age of start of treatment, compliance, age of pubic hairs appearance, breast development and menarche and how many surgeries done so far. By using SPSS calculator we will get results.

Results: Out of 27, only 5 girls (18%) have pseudo precocious puberty with the pubic hairs appear earlier but they have achieved menarche around 14 years and had regular cycles.Among these 5 girls, 4 were presented very late in endo clinic and treatment started after 1st year of life and 1 girl had very poor compliance for medicines. Remaining 23 girls have good compliance to medicines and they achieve puberty at appropriate ages and have regular menstrual cycle and breast development as well. All have undergone 1 or 2 surgeries as well.

Discussion: If CAH girls have early intervention and regular follow ups, they will have normal puberty and have regular menstrual cycles.