Introduction. Hirsutism is the exaggerated increase of terminal hair in women, developing a male model. Hirsutism is an important medical problem, not just a cosmetic defect, which currently affects about 5-15% of women. The researchers have shown that the impact of hirsutism symptoms on a woman's quality of life can be profound and can lead to psychological stress that threatens her feminine identity. Despite a large number of published works, some aspects of hirsutism are still controversial or underestimated. The treatment of hirsutism often requires a multidisciplinary approach and a variety of physical or pharmacological modalities can be used with the combination of 2 or more drugs in combination with esthetic treatment, depending on the involved etiopathogenetic mechanism.
Aim of the study. Evaluation of the molecular-genetic bases of hirsutism, the study of clinical polymorphism and the management of women with hirsutism.
Materials and methods. Online databases, meta-analyzes, scientific papers in theoretical and practical medicine were used.
Results. Following the analyses of the scientific papers we highlighted the main etiological factors of hirsutism: ovarian (93% PCOS, <1% androgendependent tumors, <1% luteoma); adrenals (<1% CAH, <1% Cushing syndrome, <1% androgen secretory tumors, 1% acromegaly) and other external factors (<1% iatrogenic, <1% androgenic drugs). The genetic approach has noted the involvement in the development of hirsutism of mutations in 5 major genes encoding important enzymes in androgen metabolism: 21-hydroxylase, P450 cytochrome oxidoreductase, aromatase, 11-beta-hydroxylase, 5 alpha reductase. The diagnosis of hirsutism is based on a detailed anamnesis, the objective exam using the Ferriman-Galway scale, seric markers, additional genetic and paraclinical tests. Ovarian suppression of androgens secretion with oral contraceptives is widely used in these women, but its efficacy is limited. One of the most effective medical therapies for hirsutism is medication with anti-androgens (spironolactone, finasteride, flutamide, etc.)
Conclusions. Hirsutism is a current medical problem that affects women's quality of life. PCOS (polycystic ovary syndrome) is the most common cause of hirsutism. The management of a woman with hirsutism requires a multidisciplinary approach - family doctor, endocrinologist, gynecologist, dermatologist. The treatment is indicated according to the ethiopathological, individualized mechanism, treatment strategies can be included with the combination of 2 or more drugs. Aesthetic and maintenance treatment is not excluded.
Key words: hirsutism, PCOS, Ferriman-Gallwey, oral contraceptives, anti-androgens

273. GENETIC ASPECTS OF FEMALE INFERTILITY

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Introduction. Female infertility is defined as the inability to conceive after one year of regular unprotected intercourse for women younger than 35 years and within 6 months for women after 35 years. One in seven couples experiences infertility or subfertility, and in 40% of cases it is because of women. Clinically, female infertility is a highly heterogeneous pathology with a complex etiology that includes environmental and genetic factors. It is difficult to assess accurately the overall magnitude of the contribution of the genetics to female infertility as most, if not all, conditions are likely to have a genetic component. Nethertheless, a significant number of infertility phenotypes have been associated with specific genetic anomalies.
Aim of the study. - This review aims to summarize current research on genetic diagnosis and genetic causes of female infertility.

Material and methods. It has been used online databases and scientific articles that contain studies of female infertility.
Results. All genetic defects can be divided into the following categories: chromosome aberrations, DNA copy number variants (micro deletions and duplications), single-gene disorders, complex conditions and epigenetic disorders. Chromosome abnormalities account for almost 60% of all spontaneous abortions, and the most common type, trisomy, is closely associated with advanced maternal age. There are 2 forms of female infertility: primary and secondary. Primary female infertility includes premature ovarian failure, polycystic ovary syndrome, endometriosis, and leiomyoma. Secondary infertility arises due to systemic or syndromic genetic defects, including developmental, endocrine, and metabolic defects. Genetic syndromes that manifest female infertility are fragile X syndrome, Noonan syndrome, sickle cell anemia, etc. Other notable conditions include disorders of sex development (SRY), reproductive dysgenesis disorders hypogonadotropic hypogonadism and Kallmann syndrome (KAl1, GNRH1, LEP) , and ambiguous genitalia an androgen insensitivity (AR). Endocrine defects comprise disruption of steroid synthesis and metabolism, and are caused by CYP17 and CYP19 mutation. Also, various metabolic defects (e.g., galactosemia) and mutation in mitochondrial energy pathway (mitochondrial DNA genes) cause toxic effects and lead to secondary female infertility.
Conclusions. The genetics of infertility is very complex and is dependent on different factors. Clearly the hope is that a greater understanding of the genetic control of infertility will bring low-risk treatment regimens that are effective and easy to administer.
Key words. Female infertility, chromosome aberrations, hypogonadotropic hypogonadism, premature ovarian failure

274. NEW COPY NUMBER VARIANTS DISCOVERED IN PATIENTS WITH OBESITY AND INTELLECTUAL DISABILITY

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Introduction. Intellectual disability (ID) is a neurodevelopment disease characterized by intellectual and adaptive impairment, defined by intelligence quotient (IQ) under 70 and can be affirmed after the age of 6. Until this age, the retard is named development delay (DD). This condition is found in 2-3% of individuals in general population, and 50% of these cases are associated with other clinical features, like pediatric obesity. The genomic study using microarray chromosomal techniques revealed in about 20% of intellectual disability patients a genetic cause of copy number variants (CNVs) type, duplication or deletion, but there is a lack of data about CNVs found in patients with ID/DD associated with obesity.
Aim of the study. To find CNVs that could be responsible for the ID/DD associated with obesity phenotype, in 36 Romanian pediatric patients, recruited from the Clinical Emergency Hospital for Children, Cluj-Napoca, Romania.
Materials and methods. We used SNP array technique, Infinum OmniExpress 24V1.2 in order to detect CNVs. Data analysis was made using Genome Studio, and the interpretation of the data was performed using UCSC data base (Decipher, ClinVar, Omim and Gene Reviews).
Results. We found relevant genetic alterations in 15 patients (42%). Several of them presented deletions and duplications that were described before in international databases, but potential pathogen CNVs not described before were also detected. Therefore, we describe a deletion inside KANSL1, the gene responsive for Koolen-De Vries syndrome, a small deletion in OTC gene, a 8p23.1 duplication in BLK gene and also a patient that presented two uniparental disomies, for chromosome 7 and 13.