به نام او که یادش آرامش بخش دلهاست

خداوند را سپاسگزاریم که توفیق برگزاری هشتمین کنگره سراسری غدد و متابولیسم کودکان و میزبانی اساتید بزرگ و همکاران گرامی فوق تخصص غدد و متابولیسم سراسر کشور را نصیب دانشگاه علوم پزشکی مازندران نمود.

لذا این امر فرصتی مغتنم برای کلیه اساتید این دانشگاه و نیز همکاران سطح استان است تا بتوانند از علم و تجربه اساتید برجسته کشور بهره مند گشته و با یاری خداوند موجبات ارتقا علمی و عملی در زمینه بیماریهای غدد و متابولیسم کودکان فراهم گردد.

استان مازندران با داشتن مناطق دیدنی و تاریخی و هوای معتدل و بهره مندی از جنگل و ساحل زیبای دریای مازندران امیدوار است که بتواند میزبان خوبی برای مهمانان گرامی بوده و این همایش بتواند در کنار فراهم کردن فرصتی مناسب برای انجام تبادلات علمی ساعات خوش و خاطره انگیزی را برای شرکت کنندگان محترم فراهم نماید.

لازم به ذکر است این کنگره با همکاری انجمن غدد کودکان و مرکز تحقیقات دیابت مازندران برگزار میگردد و برگزار کنندگان این همایش مراتب تشکر خود را از همکاری معاونت محترم آموزشی و معاونت محترم پژوهشی و مرکز آموزش مداوم دانشگاه علوم پزشکی مازندران و معاونت آموزشی و پژوهشی بیمارستان بوعلی ساری ،مرکز تحقیقات تالاسمی مازندران و کمیته تحقیقات دانشجویی دانشگاه علوم پزشکی مازندران و دفتر نهاد رهبری دانشگاه علوم پزشکی مازندران اعلام میدارد.

با امید به دیدار شما

دکتر دانیل زمانفر

دبیر علمی کنگره

ارکـــان هشتمین همایـــش سراسری غدد و متـــابولیسم کودکان

ســـاری، ۲۰ الـــی ۲۲ خـــرداد ماه ۱۳۹۴

برگزار کننده
- دانشگاه علوم پزشکی مازندران

با همکاری:
- انجمن غدد ومتابولیسم کودکان ایران
- مرکز تحقیقات دیابت دانشگاه علوم پزشکی مازندران

رئیس کنگره
دکتر قاسم جانبابایی

قائم مقام کنگره
دکتر احمدعلی عنایتی

دبیر علمی
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دبیر اجرایی
دکتر روانبخش اسماعیلی

اعضای کمیته علمی به ترتیب حروف الفبای فارسی
دکتر عذرا اخی
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دکتر حشمت مویری
سید مصطفی میر افضلی
دکتر مهین هاشمی پور
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اعضای کمیته اجرایی به ترتیب حروف الفبای فارسی
ولی اله احمدی
ابراهیم اسمعیلی
جواد اکبر زاده
شیداله آهنگری
مهدی حمیدی
حسین خراسانی
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سیدحسن سبزپوش
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سیده باجی صالحی
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دکتر مطهره خردمند، دکتر صفورا معصومی

مسئول IT
مهندس حمید رضا آذرفر

مسول دبیر خانه همایش
دکتر لیلا اسدیان

نشانی دبیرخانه
ساری میدان معلم ساختمان معاونت تحقیقات و فناوری

حمایت کنندگان هشتمین همایش سراسری غدد و متابولیسم کودکان

RISK FACTORS OF LOW PREDICTED ADULT HEIGHT IN GIRLS WITH EARLY PUBERTY

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ABSTRACT

Objective: One of the most current referrals to pediatric endocrinologists is the onset of puberty in a relatively low aged girl. In our experience frequently were seen girls with early puberty (EP) with low predicted adult height (PAH). The aim of this study was to determine the risk factors of low PAH in girls with EP.

MATERIALS AND METHODS: Girls were included in the study with onset of thelarche or pubarche at the age of 7-9.5 years. The weight, height, BMI and pubertal stage were determined. Greulich-Pyle method was used for bone age determination and Bayley-Pinneau method for prediction of adult height. Target height (TH) was calculated by mean of parental heights minus 6.5. Low PAH was defined as: PAH lower than 150 cm or lower than target zone.

RESULTS: 200 subjects had inclusion criteria for study. The mean age was 8.44±.72 years. The mean PAH and TH was 156±6.69 and 156.74±6.69 cm respectively. The 26 (13%) of subjects had PAH lower than 150 cm and 15 (7.5%) had PAH lower than target zone. Ninety one of the subjects (45.5%) had only thelarche, 17(18.5%) only pubarche and 92 (46%) both thelarche and pubarche. The risk factors of low PAH in girls with EP included: advanced pubertal stage, low birth weight, lower height SDS, advanced bone age, TH 5cm or more than PAH.

Conclusions: Although majority of girls with EP are not at risk of low PAH, but a subgroups of these girls need to medical attention.

Keywords: *predicted adult height, precocious puberty, early puberty, target height, final height*

COMPARISON OF CINNAMON EXTRACT TO METFORMIN EFFECTS UPON INSULIN RESISTANCE, APOLIPOPROTEIN B/APOLIPOPROTEIN A1 RATIO, AND BODY MASS INDEX OF OBESE ADOLESCENT GIRLS WITH POLYCYSTIC OVARY SYNDROME:A DOUBLE-BLIND, PLACEBO-CONTROLLED TRIAL

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Background: Polycystic ovary syndrome (PCOS) is one of the most common endocrinopathies, affecting 5-10% of population**.** Insulin resistance, apolipoprotein B/apolipoprotein A1 ratio, and body mass index commonly increasein obese PCO patients and are considered as the indicators of the disease. On the other hand, metformin and cinnamon are generally believed to control these endocrinopathies.

Objective: To compare the effects of cinnamon with those of the metformin on insulin resistance, apolipoprotein B/apolipoprotein A1 ratio, and body mass index of obese adolescent girls with polycystic ovary syndrome

Materials and Methods: In a prospective, double-blind, randomized, placebo-controlled clinical trial, 112 adolescent girls (12.6-17 years old) with PCOS were treated with cinnamon extract (500 mg twice daily), metformin (500 mg twice daily), or placebo, at the outpatient paediatric endocrine clinic of a university children's hospital in Tehran for 1 years.

Results: Cinnamon and metformin differed from placebo in significantly decreasing insulin resistance: both homeostasis model insulin resistance index ($p < 0.005$) and quantitative insulin sensitivity check index ($p < 0.01$), and also apolipoprotein B/apolipoprotein A1 ratio. There was no significant difference between cinnamon and metformin effects on these indexes, however, both of them slightly but significantly decreased body mass index compared to placebo ($p < 0.05$).

Conclusion: Cinnamon administration can be considered as an effective treatment for reduction of insulin resistance and weight in obese adolescent girls with polycystic ovary syndrome.

Key words: *cinnamon, metformin, polycystic ovary syndrome*

SPECIAL ASPECTS OF GROWTH HORMONE THERAPY IN CHRONIC KIDNEY DISEASE

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Growth retardation is a common complication in children with chronic kidney disease (CKD) and frequently leads to decreased adult height. Growth impairment is attributed to inadequate nutrition, metabolic acidosis, renal osteodystrophy, and disturbances in the growth hormone-insulin-like growth factor-1 (IGF-1) axis (including increased serum concentration of GH, decreased GH metabolic clearance, decreased GH receptor expression, IGF-I production and IGF activity).

Management, to prevent and correct growth impairment due to CKD, includes supportive measures in order to correct poor nutrition, metabolic acidosis, anemia and renal transplantation.

In children with persistent growth impairment (height velocity < 25th centile over a one year period, height < 3 th centile or drifting across the centiles) despite these interventions, recombinant human growth hormone therapy (rhGH) is an effective intervention, adopted since 1993.

Treatment with recombinant human growth hormone (rhGH) - an engineered form of growth hormone - in children with CKD is safe and effective, including those children on dialysis and kidney transplant recipients. It has been used to help short children with CKD attain a height more in keeping with their age group . Its use is now considered the standard care. However, There are concerns about its potential adverse effects, as well (deterioration in native kidney function, increased acute rejection in kidney transplant recipients, benign intracranial hypertension,…).

Before starting growth hormone therapy in CKD patients, abnormalities of bone and mineral metabolism and thyroid hormone should be treated. Parathyroid hormone control should be optimized as should levels of vitamin D (both 25-hydroxy vitamin D and 1,25-dihydroxy vitamin D). Blood glucose and blood sugar should be monitored.

The use of rhGH in children with CKD, including efficacy, indications, adverse effects and dosing as well a special aspects of growth hormone therapy in chronic kidney disease will be reviewed here.

Key Words: *Growth retardation ,chronic kidney disease, growth hormone*

MATURITY ONSET DIABETES OF THE YOUNG (MODY)

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ABSTRACT

Maturity Onset Diabetes of Young (MODY) refers to any of several hereditary forms of diabetes caused by mutations in an autosomal dominant gene disturbing insulin production. It is often referred to as **monogenic diabetes** to distinguish it from the more common types of diabetes, especially type 1 and type 2, which involve more complex combinations of causes involving multiple genes(i.e., **polygenic**) and environmental factors.

In MODY, there is no autoimmune destruction of β cells and no HLA association. MODY cases may make up as many as 5% of presumed type 1 and type 2 diabetes cases. The prevalence is 70 – 110 per million population. 50% of first-degree relatives will inherit the same mutation, giving them a greater than 95% lifetime risk of developing MODY themselves. For this reason correct diagnosis of this condition is important.

There are two general types of presentation: it may present with hyperglycemia, and consequent polyuria and polydipsia, or may be diagnosed simply by routine screening.

Presentation: The following characteristics suggests the possibility of a diagnosis of MODY in hyperglycemic and diabetic patients:

- Mild to moderate hyperglycemia (typically 130 – 250 mg/dl) discovered before 30 years of age.
- A first-degree relative with a similar degree of diabetes.
- Absence of positive antibodies or other autoimmunity (e.g., thyroiditis) in patient and family.
- Persistence of low insulin requirement (e.g., less than 0.5 U/kg/day) past the usual honeymoon period.
- Absence of obesity.
- Cystic kidney disease in patient or close relatives.
- Non-transient neonatal diabetes, or apparent type 1 diabetes with onset before six months of age.
- Liver adenoma or hepatocellular carcinoma in MODY type 3.
- Renal cysts, rudimentary or bicornuate uterus, vaginal aplasia, absence of vas deferens, epididymal cysts in MODY type 5.

Management: The principal treatment goals for people with MODY are the same for all known forms of diabetes. Tools of management are those for all forms of diabetes: blood testing, changes in diet, physical exercise, oral hypoglycemic agents, and insulin injections. In many cases these goals can be achieved more easily with MODY than with ordinary types 1 and 2 diabetes.

When oral hypoglycemic agents are used in MODY, the sulfonylureas remain the oral medication of first resort. When compared to patients with type 2 diabetes, MODY patients are more sensitive to sulfonylureas, such that a lower dose should be used to initiate treatment to avoid hypoglycemia.

Keywords: *Maturity Onset Diabetes of the Young (MODY), mutation, autosomal dominant gene.*

NORMAL GROWTH

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ABSTRACT

Normal growth almost always indicates absence of any major illnesses in a child. Therefore the importance of accurate evaluation and recording of growth parameters in children cannot be over emphasized. In this presentation the followings will be discussed briefly:

Pattern of normal growth

Different research methods of studying growth

Growth charts (growth attained , velocity charts)

Standard deviation and Percentiles

Characteristics of normal growth pattern

Factors affecting growth

Predilection of adult height

Shiraz study will be presented

DIABETIC NEUROPATHY IN CHILDREN

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Definition: Diabetic neuropathy (DN) is a major complication of type 1 diabetes mellitus (T1DM) with significant morbidity and mortality in adulthood. Symptomatic diabetic neuropathy is uncommon in children and adolescents but subclinical impairment has been reported in *up to 57 %* of pediatric patients.

Pathogenesis: In diabetes, a complex array of metabolic, vascular and perhaps hormonal factors shift the balance between nerve fiber damage and nerve fiber repair in favor of the former.

Implicated metabolic factors include : Accumulation of advanced glycosylation end products , Accumulation of sorbitol , Disruption of the hexosamine pathway , Disruption of the protein kinase C pathway, Activation of the poly (ADP-ribose) polymerase pathway and Increased oxidative stress .

Diagnostic methods: Diagnosis of diabetic polyneuropathy is based on: Symptom profile, Neurological examination, Quantitative sensory testing , Nerve conduction studies (NCS), Quantitative autonomic-function testing.

Screening in children and adolescents: Annual screening for the early detection of nervous system impairment should be established for all adolescents with T1DM and for diabetic children with a disease duration of more than 5 years. Testing for vibration (using a tuning fork) and pressure sensation (using a 10 g monofilament) at least annually in children older than 10 years old. Patients with abnormal or marginally abnormal results and/or objective symptoms of DN should be further examined by NCS.

Treatment and prevention: Treatment regimens are : Intensive insulin treatment , Antidepressants, Antiepileptic ,Aldose reductase inhibitors, Anti-AGE agents, ACE inhibitors, C-peptide, -lipoic acid, L-carE nitin, Protein kinase C inhibitors, Nutritional factors, Neurotrophic therapy.

Conclusion: Peripheral neuropathy is a major complication of diabetes mellitus .First signs can develop in childhood and especially during adolescence. In the presence of poor diabetic control, young people should be questioned and examined in relation to neuropathy. Patients with abnormal or marginally abnormal results and/or objective symptoms of DN should be further examined by NCS.

Key words: *Diabetic neuropathy, children*

THYROIDITIS: DEFINITIONS, CAUSES AND TREATMENTS

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ABSTRACT

Thyroiditis is aninflammation of the thyroid gland. There are several types of thyroiditis that presenting in different ways based on different etiology, so the treatment is different for each.The aim of this article is to review the clinical presentation, laboratory results, treatment, and disease outcome in literature.

The most common forms of the disease are subacute thyroiditis, is thought to have a viral origin acute suppurative thyroiditis, which is due to bacterial infection; and chronic thyroiditis, which results from autoimmune in nature

Sometimesthyroiditis could be difficult to diagnose becausethere are no symptoms unique to thyroiditis and many of the signs mimic symptoms of other diseases. Forexample subacute thyroiditis may be manifested as fever of unknown origin.

Subacute thyroiditis,as a form of thyroiditis most often presents with thyroid pain and systemic symptoms. The para clinical featuresinclude elevated ESR, elevated levels of alanine aminotransferase and alkaline phosphatase, decreased thyroid uptake of radioactive iodine and a triphasic pattern of changes in thyroid function.

Acute thyroiditisis a rare disorder caused by bacterial infection, it can be associated with anatomic abnormalities such as pyriform sinus fistula.AlthoughLeukocytosis and increased ESR are common butthe thyroid function test is most often normal.

Acute suppurative thyroiditis is a terrible clinical scenario with morbid complications andIf is left untreated it can be life threatening,

Keywords: *Subacute thyroiditis.Acute thyroiditis,chronic thyroiditis*

CONGENITAL HYPOTHYROIDISM

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ABSTRACT:

Congenital Hypothyroidism (CH) is an inadequate thyroid hormone production in newborns. It can occur because of an anatomic defect in the gland, an inborn error of thyroid metabolism, or iodine deficiency. CH is the most common treatable causes of mental retardation.

Manifestations: Infants are usually born at term or post term. Symptoms and signs include the following: Decreased activity, large fontanels, poor feeding and weight gain, small stature or poor growth, jaundice, decreased stooling or constipation, hypotonia, hoarse crying.

Screening: Early detection and treatment of CH through neonatal screening, prevents neurodevelopmental disability and optimizes developmental outcomes. The initial priority of neonatal screening for CH should be the detection of all forms of primary CH. The most sensitive test for detecting primary CH is TSH determination.

Criteria for treatment: If capillary TSH of screening is ≥ 40 mU/L, recommend starting treatment as soon as a good venous sample can be obtained, without waiting for the venous blood test result, unless venous thyroid function test (TFT) results are available on the same day. If capillary TSH concentration is <40 mU/l of whole blood, the clinician may wait for the results of venous TFT, provided that these results are available on the following day.

Treatment according to venous TFTs: If venous free T4 is below normal for age, treatment should be started immediately. If venous TSH is >20 mU/L, treatment should be started, even if FT4 concentration is normal

Assessing the cause: The thyroid gland should be imaged using either radioisotope scanning; or ultrasonography; or both.

Treatment and monitoring: L-T4 alone is medication of choice for CH. An initial L-T4 dose is 10–15 μg/kg/d. TSH concentration should be maintained in the age specific reference range; T4 or FT4 concentration should be maintained in the upper half of the age-specific reference range.

Keywords: Congenital Hypothyroidism, neonatal Screening

GOITER IN CHILDREN

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ABSTRACT:

Goiter is an enlargement of the thyroid gland. Patients with enlarged thyroids can have normal function of the gland, hypothyroidism or hyperthyroidism. Goiter may be congenital or acquired, endemic or sporadic. Congenital goiter is usually sporadic and can result from a fetal thyroxine synthetic defect or from administration of antithyroid drugs or iodides during pregnancy for the treatment of maternal thyrotoxicosis. The goiter often results from increased pituitary secretion of TSH in response to decreased circulating level of thyroid hormone. Thyroid enlargement can also result from infiltrative processes that may be inflammatory process or neoplastic disorder. Goiter in patients with graves disease and thyrotoxicosis is caused by thyrotropin receptor-stimulating antibodies (TRSAbs). Inherited defects in hormone biosynthesis are rare causes of goitrous hypothyriodism and account for only about %10-15 of the newborns with congenital hypothyroidism, another cause of goitrous hypothyroidism is resistance to thyroid hormone. Pendreds syndrome characterized by familial goiter and neurosensory deafness, iodine deficiency as a cause of congenital goiter is rare in developed countries but persists in isolated Endemic areas. The association between dietary deficiency of iodine and the prevalence of goiter or cretinism is well established. Endemic cretinism is the most serious consequence of iodine deficiency that includes 2 different but overlaping syndromes, a neurologic type and a myxedematous type. The most common cause of acquired goiter is lymphocytic thyroiditis, A rare cause of thyroiditis in children is subacute thyroiditis. Other causes include excess iodide ingestion and certain drugs. A few children with euthyriod goiters have a simple goiter, a condition of unknown cause not associated with hypothyroidism or hyperthyroidism and not caused by inflammation or neoplasm.

Key words: *Goiter, Hypothyroidism. Hyperthyriodism*

CONGENITAL DEFECT OF GLYCOSYLATION (CDG); SWEET NEWS!

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ABSTRACT

The Congenital Disorders of Glycosylation (CDG) are inherited metabolic diseases caused by defects in the biosynthesis of glycans and their attachment to proteins and lipids [1]. The glycome (defined as all glycans synthesized by an organism) is estimated to be 10–100 times larger than the proteome, so glycosylation is the most complex mechanism of molecule modification in living organisms [2] These types of metabolic diseases are rapidly growing of over 60 different disorders.

Most CDG patients have multi-organ diseases with neurological involvement. Only few of these CDG are known as non-neurological disorders [3].It is highly recommended to screen for CDG; 1) in any unexplained neurological syndrome, particularly when associated with other organ disease, 2) in any unexplained syndrome even without neurological involvement. [4]

Common characteristics are neurological abnormalities including; psychomotor retardation, seizures, hypotonia, movement disorders, stroke, microcephaly, peripheral neuropathy, abnormal myelination (like cobble stone cortical dysgenesis), and cerebellar ataxia [3,5-7]. Ophthalmological findings are also common in CDG such as convergent strabismus, retinitis pigmentosa, bilateral cataract, cloboma, glaucoma and optic nerve atrophy. [60]. Dysmorphic features like facial anomalies, inverted nipples, and subcutaneous fat pads maybe seen in some subtypes [8].

Gastrointestinal system is another common site of involvement, symptoms include; elevated hepatic enzymes, hepatomegaly, hepatic fibrosis and/or cirrhosis, steatosis, coagulopathy, hypoalbuminemia, protein-losing enteropathy, cyclic vomiting, and diarrhea [7, 9]. Sometimes endocrine abnormalities are seen in CDG, especially decreased TBG and total T4 levels, hypogonadotropic hypogonadism in combination with growth delay and decreased IGF-1 levels [10]. less common, non-CDG specific clinical features such as congenital joint contractures, ichthyosis, cardiomyopathy and abnormal rhythm, Abnormal clotting or bleeding , hearing loss, and immune systems dysfunction have been reported [5,6].

In addition, there are CDG that affect only one organ system [11] for example congenital muscle dystrophies or migration disorders of the brain [9].

A recent classification of CDGs distinguishes four major biochemical categories: [3, 11-13]

1. Defects of protein N-glycosylation (17 diseases)

2. Defects of protein O-glycosylation (8 diseases)

3. Defects of lipid glycosylation and of glycosylphosphatidylinositol (GPI) anchor glycosylation (3 diseases)

4. Defects in multiple glycosylation pathways and in other pathways (17 diseases)

The N-glycosylation pathway in the cytoplasm, endoplasmic reticulum (ER), and Golgi apparatus has four main steps: biosynthesis of lipid-linked oligosaccharide (LLO), transfer of carbohydrate chain from dolichol phosphate to theAspargine moiety of nascent polypeptide chain, remodeling of protein-bound N-glycan in the ER (basic glycosylation), and further modification of N-glycan in Golgi (final glycosylation)[5].

O-glycosylation, attachment of glycans to the hydroxyl group of threonine or serine moieties of the proteins, does not comprise processing, only assembly, which mainly occurs in the Golgi apparatus. [14]

CDG defects related to the impaired N-glycosylation are divided into two types, I and II:

CDG type I involves disrupted synthesis of the lipid linked oligosaccharide precursor (LLO) and its transfer to polypeptide chain of protein. CDG-II is characterized by impaired N-glycan processing in the end of ER and Golgi resulting in abnormal truncated species of total plasma N-glycans or serum Tf N-glycans [15-17]

Approximately 80% of patients show a type 1 pattern. With advancing biochemical techniques, the primary molecular genetic defect has been elucidated in the majority of CDG-I cases. In contrast, no comprehensive analytical or biochemical techniques are available to identify the primary defects in patients with CDG type 2. Thereby, the vast majority of these cases (80% of CDG-II patients based on EUROGLYCANET, 2010) is still classified as unsolved CDG-IIx [10].

The standard diagnostic procedure in a case of suspected CDG is the analysis of serum transferrin (Tf) either by isoelectric focusing (IEF) or by high-performance liquid chromatography (HPLC). The transferrin molecule has two complex N-glycans. These glycans are each terminated by two negatively charged molecules of sialic acid which is named tetrasialo-transferrin [3, 4,18-22] Tetrasialo-Tf is the main species in healthy individuals: in CDG-I, a decrease of tetrasialo- and an increase of disialo- and asialo-Tf are observed (type I pattern), whereas in CDG-II also monosialo- and trisialo-Tf are increased (type II pattern) [5, 11, 17]

In case with CDG type 1 pattern in IEF, the next step is the determination of two enzymes' activity: phosphomannomutase II (PMM2) and phosphomannose isomerase (PMI) in fibroblasts or leukocytes. If these results are negative, then the identification of lipid linked oligosaccharide precursor (LLO) accumulated in fibroblasts is performed. An increase of one or more N-glycan intermediates, permits to make a preliminary diagnosis that should be confirmed by further enzymatic tests and/or genetic analysis [3].

The broad application of this simple test could identify several defects of N-glycosylation. Unfortunately, similar tests unraveling defects of O-glycosylation or glycolipid glycosylation are not available, mainly because of the structural heterogeneity of O-glycans and of their tissue specific expression [24]. One of thehelpful tests in patients with CDG type II is apoC-III IEF, and should be considered complementary to selective screening. [24, 25]. Three isoforms of apoC-III can be distinguished, an increased monosialo APOC3 with decreased disialo APOC3 (APOC3-1 profile) or an increased asialo APOC3 (APOC3-0 profile) and high disialo APOC3 (APOC3-2 profile) which is seen in healthy persons [25].

Confirmatory diagnoses can only be made by molecular genetic testing in many cases. Molecular testing of single genes would not be the best option for these individuals if there is no indication of which gene may be defective. One recent advancement in the clinical diagnostic laboratory is the use of next generation sequencing (NGS) technology with gene panel testing of multiple genes involved in particular disorders, or whole exome sequencing (WES) [23, 26-28].

It is of note that not only should comprehensive mutation analysis of CDG include more than 60 genes, but also that many new genes for CDG have been discovered through WES approaches within the last years [29].

Key words: *N-Glycosylation, O-Glycosylation, Transferrin isoelectric focusing*

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ADRENOCORTICAL CARCINOMA: A CASE REPORT

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ABSTRACT

Introduction: Adrenocortical carcinoma (ACC) is a rare neoplasm with a slight predilection for female patients , with an incidence of about one case per million population. They have a bimodal peak; the first one is in the fourth and fifth decades of life and the second one in the first decade. About 60% are functional tumors that secrete hormones and present with clinical features like Cushing syndrome due to cortisone, virilizing tumor due to androgens, or feminizing tumor due to estrogens. ACC in children appears to be have differently than that in the adult patients. Virilization is more frequently seen and has a better prognosis after complete resection than in adults. The overall 5-year survival rate ranges from 16% to 38%. Recurrence, even after seemingly complete resection, is common, occurring in 23% to 85% of patients. Death usually occurs in the first 2 years.

Here in, we report a case of ACC.

Case presentation: She was of a 9-year-old girl who presented with clitoromegaly.The clinical, biochemical, histological features along with differential diagnosis are discussed. This case is presented because of the rarity, and also to highlight the importance of differentiating ACC from an adenoma particularly in pediatric patients.

CONCLUSION:ACC is an extremely rare neoplasm and particularly if it occurs in children, it is essential to differentiate it from an adrenocortical adenoma by correlating with clinical, biochemical, imaging, and histological features, because their prognoses are different.

Keywords: *Adrenocortical carcinoma, Cushing's syndrome, clitoromegaly*

A CASE IN REFRACTORY DKA: A CASE REPORT

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ABSTRACT

Baby boy 11.5 years with BW = 50kg, vomiting and loss of consciousness due to respiratory distress are referred to our center. The patient does not experience any illness. Family history of diabetes was negative. Three days before admission, the patient referred twice to the clinic and once to another hospital and was treated with serum therapy, and furazolidone.

The patient's history over the past 20 days showed polydipsia, polyuria, and loss of consciousness. In eye examination he had double Midriasis in response to light. On arrival at the emergency BS = high and ABG (PH = 6.8, Pco2 = 8, Hco3 = (non detectable), K = 4.1, Na = 130 and CBC = Nl, urinary ketones is +3. Immediately early treatment was done and the patient was received 1lit of normal saline, and then was transferred to ICU and control electrolyte and VBG was performed every two to three hours.

The treatment was done by serum therapy, insulin therapy 0.1 Iu/k/h and regulatory Electrolyte. Unfortunately, despite routine medical management Kussmall breathing and loss of consciousness continued.After that bicarbonate was started; but it didn't worked too. Finally they decided to do dialysis. After dialysis, the patient became out of DKA and PH and bicarbonate of patient gradually were corrected and the patient discharged with general goodness from hospital with prescribing insulin.

Conclusions: In diagnosis of DM and DKA, getting appropriate history and checking BS are the keys to diagnosis; and in cases of Refractory Acidosis to treatment, due to limitations in the administration of bicarbonate and the susceptibility of children to brain edema, Hemodialysis can be an effective treatment to save the patient. The developing educational programs and raising awareness of diabetes in the community, and periodic re-education of physicians about this issue and checking B.S. in patients can lead to the early diagnosis before clinical protests is severe complication or side effect.

Keywords: *Diabetic Ketoacidosis: Bicarbonate Therapy - Refractory acidosis - Hemodialysis*

REPORT OF A CASE WITH TWO INHERITED METABOLIC DISORDERS

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ABSTRACT

The patient was a 2-month-old infant who presented with weakness, abdominal distention and hepatomegaly.Her mother reported a positive history of *tyrosinemia* in patient´s *cousin*.

Initial laboratory test revealed elevated liver enzymes.According to this finding, we requested succinylacetone level in blood and then enrolled in 2-(nitro-4 trifluoromethylbenzoyl)-1, 3- cyclohexanedione (NTBC) treatment program but the patient refused the test.

One week later she was referred because of fever and gastrointestinal bleeding. Laboratory tests showed elevated international normalized ratio (INR: 6) and liver function test.

Screenings test (galactose-1-phosphate uridyl transferase (GALT) in erythrocytes and repeat succinylacetone level in blood) were request.During the length of hospital stay she was treated by antibiotic therapy and supportive care with continue NTBC.

INR level decreased to 3.5 after treatment but elevate to 4.5 four days later.

After three weeks she was admitted to hospital with sick-looking and fever.Laboratory studies detected elevated INR: 8 and low Serum phosphorus level.

At that time negative Succinylacetone; rule out the diagnosis of *tyrosinemia*. The diagnosis of galactosemia was confirmed by the assay of galactose-1-phosphate uridyltransferase in erythrocytes, showing a highly reduced activity.

She was treated with lactose-free, hydrolysate formula and NTBC was discontinued.

After treatment, the INR had decreased by a mean of 4.

Due to incomplete response to formula we request succinylacetone level in blood again which was consistent with a diagnosis of tyrosinemia type1.

This patient responded well to phosphor therapy, calcitriol, lactose-free formula, and NTBC.

The infant remained asymptomatic with complete normalization of the biochemical markers after follow-up.

Keywords: *INR, NTBC, tyrosinemia, galactosemia*

PREVALENCE AND CHARACTERISTICS OF INFANTS WITH JAUNDICE DUE TO MATERNAL DIABETES COMPARED TO INFANTS WITH UNKNOWN JAUNDICE IN GHAEM HOSPITAL IN MASHHAD DURING 2014-2007

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[1]ABSTRACT

Background: Jaundice is the common cause of hospitalization of infants in the first month after birth. Therefore, detection of risk factors associated with jaundice can affect on its process and complications. This study aimed to determine the prevalence and characteristics of diabetic mother's infants compare with infants with unknown jaundice.

Methods: In this descriptive study, of 2,800 infants with jaundice in Ghaem hospital in Mashhad during the 2007-2014, features of 59 diabetic mother's infants (case group) with 78 infants with unknown jaundice (control group) were compared. After confirming of jaundice in newborns based on pediatrician and laboratory results, a researcher made questionnaire containing maternal demographic data, maternal problems during pregnancy, mode of delivery and neonatal characteristics was completed. T-Test, Mann-Whitney, chi-square and SPSS software (19.5) were used to analyze the data.

Results: The prevalence of Jaundice due to maternal diabetes was 2.10 percent. There was a significant difference between the two groups in terms of birth weight (P=0.02), current age (P=0.003), parity (P=0.000), maternal age (P=0.000), Bilirubin (P=0.000), length of hospitalization (P=0.003), age of recovery (P=0.04), gestational age (P=0.000), mode of delivery (P=0.001), maternal problems during pregnancy (P=0.000), physical examinations (P=0.001) and jaundice diagnoses (P=0.000).

Conclusion: Diabetic mother's infants are at increased risk of high birth weight, preterm labor, maternal problems during pregnancy, cesarean, abnormal physical examinations, more jaundice and late recovery. Thus improving prenatal care, controlling maternal diabetes, management of delivery, following the neonates problems including jaundice can contribute to health promotion of these babies.

Keywords: *Diabetes, Jaundice, neonates, unknown cause*

TYPE 2 DIABETES MELLITUS IN CHILDREN AND ADOLESCENTS

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Introduction: Since the early 1990s, the incidence of type 2 diabetes has increased in children and adolescents and is linked to the rise in childhood obesity.

The following factors are associated with an increased risk for childhood onset type 2 diabetes: Obesity ,positive family history ,specific ethnic groups, female gender,conditions with insulin resistance . Childhood type 2 diabetes can present in several way: Diabetic ketoacidosis (DKA) or with only ketonuria , symptomatic presentation without ketonuria or acidosis , asymptomatic presentation

Diagnosis of diabetes mellitus in a child or adolescent is made in one of four ways : fasting plasma glucose ≥126 mg/dL . Fasting is defined as no caloric intake for at least eight hours, Symptoms of hyperglycemia and a random venous plasma glucose ≥200 mg/dL , abnormal oral glucose tolerance test (OGTT) defined as a plasma glucose ≥200 mg/dL measured two hours after a glucose load of 1.75 g/kg (maximum dose of 75 g) ,hemoglobin A1C ≥ 6.5 percent.

Both impaired fasting glucose (IFG) and impaired glucose tolerance (IGT) have been officially termed "pre-diabetes". Both categories, IFG and IGT, are risk factors for future diabetes. The diagnostic criteria used to establish these diagnoses in children are the same as those used in adults. Fasting plasma glucose 100 mg/dL to 125 mg/dL , two-hour plasma glucose 140 mg/dL to 199 mg/dL

Results: The ADA recommends screening children every three years beginning at 10 years of age or at onset of puberty (whichever comes first) if they are overweight or obese (BMI ≥85th percentile) and have two or more of the following additional risk factors : Type 2 DM in a first- or second-degree relative, member of a high-risk ethnic group: Native American, non-Hispanic black, Hispanic, Asian American, or Pacific islander , signs of insulin resistance or conditions associated with insulin resistance (eg, hypertension, dyslipidemia, acanthosis nigricans, and polycystic ovary syndrome, or small for gestational age birth weight) , maternal history of diabetes or gestational diabetes during the child's gestation. Screening for diabetes can be done by measuring hemoglobin A1C (A1C), fasting plasma glucose (FPG), or performing an oral glucose tolerance test (OGTT). Among candidates for screening who have not fasted overnight, A1C is the preferred test. Abnormal results should be confirmed either by repeating the initial test on another day, or performing a different test.

Conclusion: Treatment for type 2 diabetes is divided into nonpharmacologic, pharmacologic, and surgical therapy . Pharmacologic therapy is initiated in two settings: In asymptomatic patients who fail to achieve glycemic control three months after the initiation of lifestyle modifications, in patients who are symptomatic at presentation (eg, polyuria and polydipsia), including those with ketosis

Insulin and metformin are the only agents approved by the Food and Drug Administration (FDA) for the treatment of type 2 diabetes in children.

Keywords: *children, diabetes type 2, adolescents, obesity*

STANDARD ENTERAL FEEDING IMPROVES NUTRITIONAL STATUS COMPARED WITH HOSPITAL-PREPARED BLENDED FORMULA AMONG INTENSIVE CARE UNIT (ICU) PATIENTS

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ABSTRACT

Background: There have been great advances in enteral nutrition among critically ill patients in recent decades. However, manufactured standard enteral formula is a new material in Iran and most hospitals are still opting to use traditional hospital-prepared blended formulas.

Objective: To evaluate effects of standard enteral feeding for improving nutritional status compared with hospital-prepared blended formula among Intensive Care Unit (ICU) patients.

Materials and Methods: Forty patients participated in enteral feeding by the standard system and forty patients participated in enteral feeding by hospital-prepared blended formulas.

Results: Albumin was much increased in the standard enteral feeding group compared to hospital-prepared blended formulas ($P<0.005$). There were significant differences in results of albumin, hemoglobin (Hg) and calcium before and after feeding.

Conclusions: The macronutrients intake was elevated after standard enteral nutrition and helped patients to recover. The standard enteral formulahas more benefits than hospital-prepared blended formulas for ICU patients and medical care givers should be advised and taught to use it rather than hospital-prepared blended formula.

Keywords: *Enteral feedings, albumin, nutritional intake.*

A CATASTROPHIC ETHIOLOGY OF CHILDHOOD ENDOCRINOPATHY: CRANIOPHARYNGIOMAS

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ABSTRACT

Intruduction: Craniopharyngiomas are benign tumors of the parasellar region. These tumors may be cystic, solid, or combinations of the two. They have a bimodal age distribution and no apparent gender predilection. Patients may present with endocrinopathy or symptoms related to mass effect from the growing tumorCraniopharyngiomas are often accompanied by severe endocrine disorders. Although there is universal growth hormone deficiency(GHD), the resulting growth pattern is very heterogeneous Patients require a multidisciplinary approach during their diagnostic evaluation and subsequent to initiating therapy

discussion: Endocrinopathy should be recognized and treated with appropriate hormonal replacement. Surgery is the first-line therapy for most paients. The specific surgical approach must be tailored to the specific clinical situation and depends on the patient age, endocrine status, and the geometry and consistency of the tumor. Whereas most solid tumors will require craniotomy or transsphenoidal surgery, some cystic tumors may be adequately managed with intracavitary therapies. Subtotally resected or residual tumors often require adjuvant radiation therapy or radiosugery. Long-term multidisciplinary follow-up is necessary for all patients

Conclusion: The endocrine morbidity could develop in most children with craniopharyngioma before and after the operation and should be managed properly. Although all treated patients benefit from GH therapy, further studies are necessary to find out the possible mechanism of growth regulation in normally growing children, despite GH deficient.

Key word: *craniopharyngioma,endocrinopathy,childhood*

PRECOCIOUS TRUE PUBERTY SECONDARY TO A SUPRASELLAR ARACHNOID CYST: A CASE REPORT AND MR CHARACTERISTICS OF COMMON SUPRASELLAR CYSTIC LESIONS

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Introduction: Arachnoid cysts are intra-arachnoid collections of cerebrospinal fluid (CSF).9–11% of these cysts are located in the suprasellarregion.The exact incidence of true precocious puberty with an SSAC is unknown, and it is an uncommon presentation with only sporadic reports in the literature.

Case report: An 11-year-old boy presented with a history of an abnormal increase in the size of his external genitalia and a change in his voice over a period of 2 years.He exhibited mild macrocephaly, which manifested primarily as a prominent forehead. His sexual development, which was determined by his pubic hair and testicular volume, was classified as Tanner stageV.An endocrinological profile revealed the following values: normal levels of luteinizing hormone (2.92 IU/L), follicle-stimulating hormone (1.61 IU/L) and thyroid stimulating hormone (1.71 mL U/L); increased levels of testosterone (32.93 nmol/L); and decreased levels of free thyroxine (8.84 pmol/L).

Non-enhanced magnetic resonance imaging (MRI) of the patient's brain (GE 1.5 T) revealed a large suprasellar cyst that measured $3 \times 4 \times 4$ cm^3.The data indicated a mass effect, which involved stretching and anterior displacement of the optic chiasm andpituitary stalk.

Discussion: The majority of patients with suprasellar arachnoid cysts that have been described in the literature are children and.The pathogenesis of precocious puberty with an SSAC is controversial.The intact anterior hypothalamus, in the absence of inhibitory influences, leads to an increased level of pituitary function.

Conclusion: Although the most common tumor in precious puberty is a hamartoma, an arachnoid cyst should be considered in the differential diagnosis. Patients with suprasellar arachnoid cysts should be observed over long periods of time to monitor their growth and pubertal development

Key word: *Precocious puberty; Suprasellar arachnoid cyst, diagnosis*

EVALUATION THE RELATIONSHIP BETWEEN OBESITY AND HYPERTENSION IN PRIMARY SCHOOL CHILDREN OF TORBATHEYDARIEH IN 2013

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Background: Several studies in children have shown that there is a meaningful relationship between blood pressure and Body Mass Index (BMI).

Objective: current study was conducted to investigate the association between systolic and diastolic blood pressure with BMI in primary school children of TorbatHeydarieh in 2012-2013

Materials and methods: This study was conducted in TorbatHeydarieh(between2012-201). The participants were 1750 primary school children selected through systematic cluster sampling. Their heights and weights were measured, analyzed and the mean was considered. The data obtained from statistical analyses was analyzed based on research goal.

Results: Out of 1750 participants (920 boys and 830 girls) 166 were not fat, 88 subjects were fat, 1592 had a normal blood pressure, 158 had a high blood pressure; 115 out of 158 hypertensivepeople were not fat. Relative frequency of high blood pressure in non-fat was 4.45% and in fat people was 83.2%there wasa significant relationship between fatness and high blood pressure.There was also asignificant relationship between systolic and diastolic blood pressure and BMI in both genders among all participants ($p<0.05$)

There was a meaningful relationship between age and systolic blood pressure in patient aged 7 -11 years old and diastolic blood pressure was meaningfully related to BMI in patient aged 9-11 years old

Conclusion: According to the mutual effects of fatness and hypertension it is recommended that child nutrition programs should be further considered and screening program for fatness should be implemented among schoolage children that lead to prevention and early detection or treatment of the disease.

Keywords: *hypertion-obesity-children*

OBESITY IN CHILDREN

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ABSTRACT

Context: Obesity is associated with a higher energy consumption than to the other causes; the most important metabolic factor, lifestyle ,diet and physical activity noted that each of these factors can be influenced by heredity fall.Fed exclusively with breast milk in reducing the risk of obesity has a significant effect .Children who eat non-nutritious food, get more fat and salt than other children ,that predispose them to chronic diseases such as obesity, high blood pressure,diabetes, cardiovascular disease and cancer later in their life , because eating habits are formed in childhood and in adulthood affects a person's actions and attitudes.Obese children and adolescents are not only at risk of complications such as high blood pressure, blood lipid disorders,type 2 diabetes, but also because of poor diet to lack of micronutrients and also due to the consumption of foods with high calories,The nutritional value is low. The purpose of this study was to review the relationship between diet and obesity disease in children in the future.

Evidence Acquisition: In this review, a literature search in pubmed, magiran and google scholar were provided.

Results: Several research findings suggest the role of various factors in obesity, especially after childhood is high or low birth weight, type of milk and duration of breastfeeding, including requirements. These results show the importance of proper nutrition in health and its effect on the prevention of the complications of obesity are clear. Obesity increases the risk of complications such as atherosclerosis, hyperinsulinemia, high blood pressure and psychological problems in young people.

Conclusion: To improve nutrition education to families in different ways to reduce the prevalence of obesity and ultimately improve public health. Given the increasing prevalence of obesity in children and adolescents and the prevalence of iron deficiency anemia and other micronutrient in obese children seem to be the screening of iron-deficiency anemia in children and adolescents should be done with a high pitch.The results of the studies show a higher prevalence of risk factors for cardiovascular disease in obese children, and the prevention and control of obesity in children soon after the beginning of time a child is stressed.

Keywords: *obesity, diabetes, diet*

POLYCYSTIC OVARY SYNDROME IN YOUTH

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ABSTRACT

PCOSis a heterogeneous syndrome of unexplained chronic hyperandrogenism and oligo-anovulation and the most common cause of chronic anovulation associated with hyperandrogenic state. Its incidenceis about 5-10% in reproductive-age women. Diagnosis is made by excluding other hyperandrogenic disorders (e.g., nonclassic and classic adrenal hyperplasia, androgen secreting tumors, hyperprolactinemia) in women with chronic anovulation and androgen excess.

Major morbidities are related to reproductive and cardiovascular systems. The most important reproductive morbiditiesconsisting infertility,irregular uterine bleeding, increased pregnancy loss and higher risk of endometrialcancer than non PCOS people.Cardiovascular risks arelinked to insulin resistance and common occurrence of obesity, although it also occurs in non-obese women with PCOS.

PCOS is considered to be a heterogeneous disorder withmultifactorial causes and it appears to account for 70% of the variance in pathogenesis. Both heritable and non-heritable factors contribute to arise it.So a positive family history of chronic anovulation andandrogen excess increases the risk for occurring PCOS.

There are many evidences that risk factors for PCOS can be recognized in childhood. Congenital virilizing disorders; above average or low birth weight for gestational age; premature adrenarche, particularly exaggerated adrenarche; atypical sexual precocity; or intractable obesity with acanthosis nigricans, metabolic syndrome, and pseudo-Cushing syndrome or pseudo-acromegaly in early childhood have been identified as independent prepubertal risk factors for the development of PCOS. During adolescence, PCOS may masquerade as physiological adolescent anovulation. Asymptomatic adolescents with a polycystic ovary occasionally (8%) have subclinical PCOS but often (42%) have a subclinical PCOS type of ovarian dysfunction, the prognosis for which is unclear.

Identifying children at risk for PCOS offers the prospect of eventually preventing some of long-term complications associated with this syndrome once our understanding of the basis of the disorder improves.

Keywords: *PCOS, Adolescent, Children, Youth*

CARBAGLU® AND THE TREATMENT OF HYPERAMMONAEMIA

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ABSTRACT

Hyperammonaemia is defined as plasma ammonia concentrations above the normal range. It should be noted that ammonia levels depend on many factors including age of the patients and assay.

The brain is the main organ affected by hyperammonaemia. Hyperammonaemia causes reversible and irreversible metabolic and neurotransmitter disturbances leading to severe brain toxicity .Increased ammonia levels lead to increase glutamine levels, which in turn lead to increase intracranial pressure. Increased brain glutamine causes increasing of membrane permeability and astrocyte swelling. The metabolic breakdown of proteins and amino acids releases nitrogen in the form of ammonia. Since ammonia is a neurotoxin, it must be converted to a relatively non-toxic form and excreted from the body to prevent damage to the brain. The hepatic urea cycle is the primary pathway for the excretion of ammonia, by converting it to urea which is easily removed by the kidneys. Since ammonia is toxic mainly to the brain, most of the signs and symptoms of hyperammonaemia are neurological manifestations. Urea cycle disorders are due to deficiencies in the liver enzymes involved in the metabolism of excess nitrogen.

A deficiency in any of these can result in a failure of the urea cycle. The primary goal in the treatment of NAGS deficiency is to rapidly and effectively reduce acute hyperammonaemia to avoid the associated and irreversible complications of the CNS. Untreated NAGS deficiency might be fatal because of the severe toxicity associated with hyperammonaemia and glutinaemia. Emergency treatment involves:

1. Low protein diet
2. High calorie supplements
3. IV glucose and arginine hydrochloride
4. Ammonia scavengers (eg sodium benzoate, sodium phenylacetate or sodium phenylbutyrate)
5. Haemodialysis as rescue therapy
6. Carbaglu® to restore urea cycle activity

Keywords: *Carbaglu, hyperammonaemia, metabolic disturbances*

IMPORTANCE OF GROWTH MONITORING AND EARLY REFERRAL OF SHORT STATURE

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ABSTRACT

• During the first two years of life a rapid growth occurs. From 2 years until about 10 years of age, the child will grow at a steady rate of approximately 5 to 7 cm a year. The growth spurt that goes along with adolescence begins at about age 11 in girls and 13 in boys. This pubertal growth spurt usually lasts 2 years and is accompanied by sexual development.

• Growth ceases between 16 and 18 years of age, when the growing is ended by fusing of the bones. During puberty, sex steroids and growth hormone both play a role in the pubertal growth spurt. Sex hormones are involved in the process of growth plate fusion which stops linear growth.

• Somatic growth and maturation are influenced by several intrinsic and extrinsic factors that act independently, or in concert, to modify an individual's growth potential.

• Differences in growth and development vary as a function of sex and ethnic origin.

• Growth hormone regulates childhood growth, but also other hormones have impact such as thyroid hormone, insulin and sex hormones.

• A few compelling data implicate excessive exercise as causal in the shorter stature of some pubertal athlete.

• Worldwide, the single most common cause of growth disturbances is poverty-related malnutrition.

• Growth hormone deficiency causes numerous metabolic and physiological dysfunctions in children and adults, including the reduction in linear growth in children and an increase in visceral fat and decreased muscle mass and bone density in adults. Growth retardation is a relatively early sign of poor health. In developing countries, growth monitoring is primarily aimed at detecting malnutrition. In industrialised countries, the focus of growth monitoring is on early detection of growth disorders such as Turner syndrome and growth hormone deficiency (GHD

Keywords: *short stature, Growth hormone, deficiency*

INSULIN ASPART IN THE MANAGEMENT OF TYPE 1 DM

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ABSTRACT

Insulin aspart is a biosynthetically modified analogue of human insulin. A single proline amino acid at position 28 of the insulin B-chain has been replaced with an aspartic acid residue. In all other respects, insulin aspart and human insulin are structurally identical. At all doses the early pharmacodynamic activity of insulin aspart is higher than with regular human insulin. Insulin aspart provides comparable overall pharmacodynamic activity to regular human insulin. Insulin aspart has a faster onset and a shorter duration of action than soluble human insulin. Glucose excursions are similar when insulin aspart is administered after meals and human insulin before meals. Insulin aspart has a significantly lower postprandial glucose increment than human insulin in T1DM. Insulin aspart has faster onset of action and shorter duration of action than human insulin leading to better postprandial glucose control. Insulin aspart has better HbA_{1c} control compared to human insulin in basal-bolus therapy .Insulin aspart has a lower rate of hypoglycaemia (especially nocturnal) compared to human insulin. Rates of symptomatic and nocturnal hypoglycaemia were significantly lower with insulin aspart and lispro than glulisine. The percentage of patients with significant ketosis and/or risk level for impending diabetic ketoacidosis was higher for insulin glulisine compared to aspart and lispro.In both adults and children with inadequately controlled type 1 diabetes, sensor augmented pump therapy resulted in significant improvement in glycated hemoglobin levels, as compared with injection therapy. Postprandial insulin aspart is equally effective and safe compared with preprandial human insulin.More patients achieve HbA_{1c} targets with insulin aspart compared to insulin lispro with a significantly lower daily insulin dose.

Keywords: *type 1 DM, Insulin aspart, HbA_{1c}*

THE ETIOLOGIES OF SHORT STATURE AMONG CHILDREN AND ADOLESCENTS REFERRED TO PEDIATRIC ENDOCRINOLOGY CLINIC OF KASHAN UNIVERSITY OF MEDICAL SCIENCES

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ABSTRACT:

Introduction: Short stature is one of the most common causes of referral to the endocrinology and pediatric clinics. Short stature in most cases is due to physiological causes like as familial or constitutional short stature, but it may be due to some curable and important diseases. In this study we investigate the causes of short stature.

Material and Method: Every child under 18 years old whom had height below 3rd percentile of growth charts, referred to Kashan medical university endocrinology clinic, included to the study as her/him parent wished. The height and weight of the children and their parents were measured and physical examination was done in order to find out the cause of short stature. For all children a set of routine lab data performed. The left hand x-ray of each child was taken to identify the bone age and evidence of chondrodysplasia. Growth hormone stimulation test applied if there was any indication for this test.

Result: 363 short stature child and adolescent (173 male, 190 female) were studied. The more common causes of short stature included: familial short stature 39.9% (112cases), constitutional growth delay 26.4% (96cases), growth hormone deficiency 7.96% (29cases). The less common causes of short stature included: hypopitutarism , hypothyroidism , achondroplasia ,hypochondroplasia, adison, cancer, chronic hepatitis, congenital ichtiosis, cystic fibrosis, cystinosis, Down syndrome and etc.

Conclusion: The short stature in children has a wide spectrum of causes that early and perfect diagnosis can lead to better treatment.

Key words: *Short stature, Growth, Constitutional growth delay, Familial Short stature, Children*

GONADOTROPIN-RELEASING HORMONE AGONIST THERAPY AND OBESITY IN GIRLS: IS A CONCERN?

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ABSTRACT

Objective: To investigate the effect of gonadotropin-releasing hormone agonists (GnRHa) therapy on body mass index (BMI) in girls with central precocious puberty (CPP).

MATERIALS AND METHODS: The diagnosis of PP in girls was made based upon the onset of secondary sexual characteristics before 8 yr of age in or menses before 9 yr of age. Patients were included in the study if 1) CPP has been proved with clinical and laboratory criteria and 2) good compliance with study protocol. The weight, height, BMI and pubertal stage were determined before treatment, and 6 and 12 month after. Marshall-Tanner method was used for sexual maturation staging. Intramuscular injection of GnRH agonist (Triptorelin) was administered for the patients with rapidly progressive forms of CPP. Patients with slowly progressive forms of CPP were considered as control group.

Results: 110 subjects had inclusion criteria for study, 46 (48.1%) patients as cases and 62.85)4%) patients as controls. The mean age at initial visit was 7.4 31.0±6years (7.89±1.15years in cases and7.10.81±6years in controls, P value <)0.0001.The weight, height, BMI, and the prevalence of obesity and overweight was not significantly different in cases compared with controls. The BMI had significant increase in cases and controls at 6 and12 months compared with onset of treatment (p<0.0001). The BMI SDS in cases and controls was not significantly different at 6th and12th months compared with onset of treatment (p=0.752, p=0. 938 respectively) .The prevalence of obesity was not different between cases and controls at first and 6 &12 months of therapy (p=0.11, p=0.068, p=0.052 respectively) .

Conclusions: GnRHa therapy has not effect on body mass index and the prevalence of obesity does not change with GnRHa therapy.

Keywords: *GnRHa therapy, precocious puberty, obesity*

ADRENAL INSUFFICIENCY

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ABSTRACT

Definition: Adrenal insufficiency is a condition in which the adrenal glands do not produce adequate amounts of steroid hormones, primarily cortisol; but may also include impaired production of aldosterone.

Types: Primary adrenal insufficiency is due to impairment of the adrenal glands(Addison's disease, idiopathic, congenital adrenal hyperplasia or an adenoma),secondary adrenal insufficiency is caused by impairment of the pituitary gland (exogenous steroid use, pituitary adenoma) ,and tertiary adrenal insufficiency is due to hypothalamic disease.

Signs and symptoms:Include hypoglycemia, dehydration, weight loss, disorientation, weakness, tiredness, dizziness, low blood pressure, orthostatic hypotension, cardiovascular collapse, muscleaches, nausea, vomiting, and diarrhea. These problems may develop gradually and insidiously.

Diagnosis: The best diagnostic tool is the ACTH stimulation test; however, if a patient is suspected to be suffering from an acute adrenal crisis, immediate treatment with IV corticosteroids is imperative and should not be delayed for any testing. If ACTH test not performed during crisis then labs to be run should include: random cortisol, serum ACTH, aldosterone, renin, potassium and sodium. A CT of the adrenal glandsand MRI of the pituitary can be used.

Treatment: *Adrenal crisis*: Isotonic sodium chloride solution 450 mL/m^2 or 20 mL/kg is given over the first hour, a second 20-mL/kg bolus of isotonic sodium chloride solution may be given. Follow this with continuous infusion of 3200 mL/m^2/d. Stress doses of hydrocortisone 50-75 mg/m^2 given intravenously, followed by 50-75 mg/m^2/d divided in 4 intravenous doses.*Long-Term therapy*: In a child with adrenal insufficiency long-term glucocorticoid replacement must be balanced between the need to prevent symptoms of adrenal insufficiency and the need to allow the child to grow at a normal rate. The range for hydrocortisone is 7-20 mg/m^2/d given orally in 2 or 3 divided doses. Patients with primary adrenal insufficiency who also have mineralocorticoid deficiency require fludrocortisone at 0.1-0.2 mg/d.

Keywords: *Adrenal gland, Insufficiency, Treatment, clinical signs*

EPILEPTIC ENCEPHALOPATHY AND METABOLIC DISEASE

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ABSTRACT

Epileptic encephalopathy can be induced by inborn metabolic defects that may be rare individually, but in aggregate represent a substantial clinical portion of child neurology. These may present with various epilepsy phenotypes including refractory neonatal seizures, early myoclonic encephalopathy, early infantile epileptic encephalopathy, infantile spasms, and generalized epilepsies which in particular include myoclonic seizures. There are varying degrees of treatability, but the outcome if untreated can often be catastrophic. The importance of early recognition cannot be overemphasized. Selected diseases are organized by the defective molecule or mechanism and categorized as small molecule disorders (involving amino and organic acids, fatty acids, neurotransmitters, urea cycle, vitamins and cofactors, and mitochondria) and large molecule disorders (including lysosomal storage disorders, peroxisomal disorders, glycosylation disorders, and leukodystrophies). Details including key clinical features, salient electrophysiological and neuroradiological findings, biochemical findings, and treatment options are summarized for prominent disorders in each category. Inherited metabolic epilepsies are disorders that, while individually rare, are in aggregate a substantial clinical portion of child neurology, as well as a complex field of knowledge for physicians, investigators, and students to tackle. A subset of these disorders can lead to the development of epileptic encephalopathy, that is, a brain disturbance due to highly active clinical or electrographic ictal activity. The epileptologist may view these from the viewpoint of syndromic phenotypes such as early myoclonic encephalopathy, early infantile epileptic encephalopathy, infantile spasms, and myoclonic epilepsies. They have various degrees of treatability at present, with some requiring prompt diagnosis and intervention to avoid otherwise catastrophic outcomes.

Keywords: *Epileptic encephalopathy, inborn metabolic defects, child neurology*

MOST COMMON FUNGAL INFECTION IN ENDOCRINOPATHIES_DIAGNOSTIC AND THERAPEUTIC PROBLEMS.

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Objective: The frequency of mycoses in endocrinopathies has increased during recent years. Some of fungi become colonized on surface skin and mucous membranes and may invade the internal organs through blood vessels become life threatening. The risk of mycoses is determined both by pathogenic properties of fungi and host characteristics. Endocrine dysfunction (e.g. AIDS, tumors, diabetes and other hormone dysfunction) are major risks of mycosis. Fungal infections can manifest from asymptomatic colonization to invasive and their significance is underestimated and proper diagnostic procedures are not conducted. The aim of this article is to summarize the current knowledge on diagnostic and therapeutic problems of fungal infections in endocrinopathies.

Method: A literature and institutional review was completed for fungal infection in endocrinopathies over the past 25 years including common agents, difficult diagnosis, treatment problems and outcome.

Results: The Most common type of mycoses in endocrine dysfunction include infections of the oral cavity, gastrointestinal tract, skin and it's appendages, Urogenital system, blood system (fungemia), central nervous system and disseminated. The most common pathogenic fungi in this group are Candida spp., Aspergillus spp., Coccidioides spp., and Cryptococcus spp. Diagnosis and treatment of fungal infection particularly in systemic type in endocrinopathies is major problem. Because conventional diagnosis of these infections based on culture of blood or offending organism from multiple sites, often delays therapy. When antifungal therapy is instituted, inappropriate dosing seems quite common.

Conclusions: High incidence of mycoses in endocrinopathies, possible severe complications and increasing number of species resistant to standard antifungal agents, considering whether the mycological examination should be introduced into the panel of methods evaluating clinical condition of endocrinopathies both in outpatient clinics and hospital departments. The diagnostic procedures require extension. It is worth mentioning that it can be extremely helpful in controlling systemic infections.

Key words: *Endocrine dysfunction, Fungal infection, Mycoses, Diagnosis, Treatment, Antifungal drugs*

INTERPRETATION OF METABOLIC LAB. RESULTS

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In clinical conditions with suspicion to metabolic disorders (such as convulsion, developmental delay, hypoglycemia, metabolic acidosis and …), checking of ABG and blood ammonia for differential diagnosis is helpful:

In UCD[1], elevation of blood ammonia (usually above 300μmol/L in severe form) without metabolic acidosis exists.

Increased level of blood citrulline, glutamic acid, aspartic acid, alanine and urine level of orotic acid and uracil is useful for diagnosis.

In aminoacidopathies, ABG[2] and ammonia level is usually normal:

2. a) PKU[3]: blood level elevation of phenylalanine (usually above than 10mg/dl) with positive urine metabolites (phenylpyruvate phenylacetate) is diagnostic.

2. b) Tyrosinemia: high level of tyrosine with HPLC[4] method in blood with urine elevation of succcinylacetone- N, acetyltyrosine- 4 HPPA[5]- 4 HPLA[6]- 4 HPAA[7] (detected by GCMS[8] method) is seen.

Organic acidemia:

3. a) Branched chain aminoacid disorders (ßKT[9], PA[10], MMA[11], …):

Detection of elevated acyl carnitine profiles (such as C3- C5, …) in blood by MS/MS[12] method and urine increased level of organic acids (3-hydroxy propionate, methylmalonate, isovalerylglycine…) is necessary.

1 Urea cycle disease
2 Arterial blood gas
3 Phenylketonuria
4 High performance liquid chromatography
5 4 Hydroxy phenyl pyrovic acid
6 4 Hydroxy phenyl lactice acid
7 4 Hydroxy phenyl acetice acid
8 Gas chromatography mass spectrometry
9 Beta ketothiolase
10 PA- Propionic acidemia
11 MMA- Methylmalonic acidemia
12 Tandem mass spectrometry

3. b) FAOD[13]: assessment of acyl carnitine profiles (C_0- C_2- C_4- C_8- C_{16}- C_{18}, …) with MS/MS method is diagnostic.

Also, nonketotic dicarboxylic aciduria is seen in urine analysis with GCMS method.

3. c) Mitochondrial disorders: there is not any specific finding in acyl carnitine profiles for these disorders. High blood level of lactate and lactate/ pyrovate ratio with increased urine excretion of lactate- 3 hydroxybutyrate, acetoacetate, fumarate, succinate and malate are indicative for mitochondrial disorders.

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13 Fatty/ acid oxidation disorders

EVALUATION OF CLINICAL MANIFESTATION IN DIABETIC KETOACIDOSIS

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Introduction and Objectives: DKA is the end result of the metabolic abnormalities resultingfrom a severe deficiency of insulin or insulin effectiveness. DKA occurs in 20-40% of children with new-onsetdiabetes and in children with known diabetes who omit insulindoses or who do not successfully manage an intercurrent illness.The aim of this study was to evaluate patient with DKAadmitted in pediatrichospitals ofKhorramAbad.

Materials and Methods: Thisstudy was done cross – sectional. All patients that hospitalized with DKAwere evaluated in 2010-2014.Studied variables include sex, age, clinical and Para clinical data and period of hospitalization. Data collected by questionnaire and analyzed by Spss program.

Results: Out of 50patients with DKA 68% were girls and 32% boys. 8% of patients was less than 5 years, 53%was 6-10 years and 39% 10-14 years. Theaverage of age was 8/7 ±3.5. 55% was urban and 45%were rural.35% hospitalized 2 times. Clinical signs included: 45% polyuria and polydipsia, 23%nocturnal enuresis, 28% decrease of consciousness, 25% abdominal pain, 20% upper respiratory infection, 6% headache and vomiting. Average of blood suger in admission time was 375±85 .Gulucosuria 3+ in 78% and Ketonuria 1+ in 74% .period of hospitalization was 7-12 days.

Conclusion: inThisstudy 68% were girls, 53%was 6-10 years, 28% decrease of consciousness. Considering that diabetic mellitus is the most common endocrine metabolic disorder of childhood, parents must be educated.

Keywords: *children, diabetic ketoacidosis*

THE STUDY OF DIMENHYDRINATE ANTIHISTAMINE`S EFFECTS ON LIVER`S TISSUE AND ENZYMES IN MALE RAT

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ABSTRACT

Objectives: Dimenhydramine is a drug with anti-gastric reflux and anti-nausea effects that usually used in trip for sensitive persons. This drug absorbed easily from oral usage and during 24 hours excreted from kidneys and it has hepatic metabolism.

In this study we checked the distractive effects of dimenhydramine antihistamine on liver`s tissue and enzymes thereinafter excessive prescription.

Material and method: We used 40 male wistar adult rats whit 250+20 weighting that grouped in four groups. The control group was without treatment and the other fed with 2.5, 5, 7.5 mg/kg of dimenhydramine doses daily within 28 days. After the examination procedure, rats became unconscious with ether, blood sampling and liver`s biopsy has been done and sent to laboratory for more study.

Result: Comparing the enzyme levels of ALT, AST and ALP in the treatment and control groups, it represented that there was a rise in the enzyme levels as a result of liver damage caused by taking dimenhydramine antihistamine. Observing the microscopic slides of the liver tissue of the treatment group, it suggests sinusoid destruction, the loss of bile ducts, irregular placement of adjacent cells and the absence of Kupffer cells, which in turn confirms the negative effect of this drug on liver tissue.

Conclusions: Like other medications, dimenhydramine can exert adversary effects as well as positive effects. Since one of the tissues which are susceptible to this effect is liver, we must bear in mind when it comes to prescribing this medication.

Keywords: *Liver, dimenhydramine, antihistamine*

ROLE OF LABORATORY METHODS TO IDENTIFY THE DISORDERS ASSOCIATED WITH METABOLIC ERRORS

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ABSTRACT:

In general, metabolic disorders are divided intosix categories: aminoacid, carbohydrate, fatty acid, organic acid disorders, mitochondrial, lysosomal and peroxysomal functional disorders.

The primary line of laboratory evaluation starts with following tests: **Blood:** CBC, electrolytes, ammonia, uric acid, gas, lactate, and pyruvate; **Urine:** odor, pH, ketone, and reducing substances; **CSF:** lactate, pyruvate, glucose.

Following collecting data from mentioned above tests, the second line of investigation will befinding abnormal values of certain metabolites related to any of 6 categories of disorders. At this stage, HPLC and Tandem Mass Spectrometryare done followed by GC-Mass Spectrometry, as a confirmatory approach to initial findings. Eventually, an enzyme assay helps pinpoint disorder. Final laboratory diagnosis can be shaped by performing nucleic acid analysis, as complement ofthe second line of investigation cycle.

Conclusion: Necessity of Improvement Our Approach to Laboratory Diagnosis

For laboratory investigation of metabolic disorders In Iran, nationwide strategies are now defined and developed; however there are still some barriers in this way and among them following challenges that need to be certainly addressed: attention to major centers performing laboratory tests dealing with metabolic disorders; training laboratory specialists and technicians, and medical specialists of metabolic disorders to become familiar with latest findings and discoveries in the field; connection with distinguished and accredited international centers to obtain information at the edge of science.

Key words: *mitochondrial - Tandem Mass Spectrometry - metabolic disorders - nucleic acid analysis*

AUDITING NURSING CARES REGARDING NEONATE OF DIABETIC MOTHERS AT BABOL UNIVERSITY OF MEDICAL SCIENCES SELECTED HOSPITAL IN 2014

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ABSTRACT

Background: Nursing cares regarding neonate of diabetic mother, is a key factor in increasing of chance of life of this neonates.

Objectives: The aim of this study was to investigate nursing care conformity rate with existing standards, regarding neonate of diabetic mother.

Materials and Methods: In this descriptive study, 400 nursing care regarding to neonate of diabetic mother were observed. Data collected using 3 checklists included standard cares regarding to neonate of diabetic mother, which was developed according to reviewing literatures and existing standards. Content validity and inter rater coefficient reliability (ICC=0.95 in neonatal, NICU and operation room wards, in maternity ICC=0.98) were calculated for checklists. Data collection method was observation and recording items of checklists. Sampling was in two methods: Event sampling and time sampling which last 4 months. Data analyzed by statistical methods and $P < 0.05$.

Results: Conformity rate of nursing cares regarding to neonate of diabetic mother in Neonatal and NICU wards of selected hospitals , maternity and operation room were 85.1%, 79% and 62.6% respectively.

Conclusion: Nursing cares regarding neonate of diabetic mother in operation room and maternity are far from standards, which can attribute to lack of familiarity of health providers and lack of training.

Keywords: *Auditing, Neonatal intensive cares, infant of diabetic mother*

ABETALIPOPROTEINEMIA: A CASE REPORT AND LITERATURE REVIEW

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ABSTRACT

Introduction: Abetalipoproteinemia (ABL) is a very rare (<1:1000,000) autosomal recessive metabolic disorder that prevents from normal absorption of dietary fats. Mutations in the MTP gene encoding the MTP (Microsomal triglyceride transfer protein) cause the disease .This protein has an important role in the transfer of lipids onto apo B and it's absence compromises the transport of absorbed fats into the lymphatic system and the general circulation. The disease is usually revealed during early childhood by steatorrhea and failure to thrive. In later childhood, ataxia and retinitis pigmentosa will be appeared.

Case presentation: Here, a 19-month-old patient with Abetalipoproteinemiais presented. He was pale, and had a bulging abdomen. His weight and height were below the 5th percentile,The child's appearance and severe failure to thrive pushed the physicians to look for possible many metabolic abnormalities. Although, so many additional diagnostic and laboratory tests were performed for him at the time of the visits, but nobody could be identified his problem. when his mother reported that he had foul-smelling stool from the first month of life, Microscopic examination of the stool revealed steatorrheaand confirmed this fact .In this regard extremely low plasma lipids levels, absence of beta-band in lipoprotein electrophoresis and hematologic manifestations of ABL include acanthocytosis were detected.

Conclusion: If your patient have any symptoms of ABL,such as abnormal growth patterns and fatty and frothy stools, you can think to abetalipoproteinemia. Remember ABL is treatable, but treatment delays can have lasting effects.

Key Word: *Abetalipoproteinemia, steatorrhea;failure to thrive, APO B*

MANAGING DIABETES WITH NANODIAGNOSTICS, NANOTHERAPUTICS, AND NANOVACCINES

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ABSTRACT

Nanobiotechnological approaches would be potent candidates for improving the care of patients with diabetes. Various nanoparticles are being appliedfor imaging contrast agents to help in the early diagnosis of type 1 diabetes. Nanobiosensors are being applied in implantable forms that enable rapid real-time monitoring of blood glucose levels, and these nanodevices would also be used as the basis ofsmart glucose-responsive nanoparticles for mimicking the body's physiological needs for insulin. However,"Nanobiotechnology"would be used for non-invasive delivery of insulin and engineering more effective nanovaccines for cell and gene therapies in diabetes. Here, we introduced the current state of nanomedicine approaches and explainimportantways for their applicationsindiabetes medicine.

Keywords: *Diabetes, Nanodiagnostics, Nanotheraputics, Nanovaccines, Nanomedicine*

THE EFFECT OF AEROBIC EXERCISES AND REZVIN SUPPLEMENT ON SERUM LEVEL OF SIRTUIN1 IN WOMEN WITH TYPE 2 DIABETES

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intruduction: Aerobic exercise is a training in which in using the function of energy generating organs, increases that leads to the increment in the cardio-spiratory endurance and includes continuous activities. Physical activities and herbal supplements were considered as a basis for the treatment of the diabetes. The purpose of the present study was to determine the effectsof aerobic exercises and Rezvin supplement on serum level of Sirtuin -1 in women with type 2 diabetes.

Methods and materials: in this semi-experimental study with pretest -posttest design, 27 women with type 2 diabetes (age 27/945/32± and body mass index: 28/665/3 ± kg/m^2) based on availability criteria, were randomly assigned to two experimental groups (N=18) and one control group (N=9). One of the experimental groups participated in 8 weeks of aerobic exercise program three days a week with maximum heartbeat of 65-75% and the second experimental group in addition to the exercises, received daily Rezvin supplement containing 400mg resveratrol for 60 days while, during this period the control group spent their normal life without any intervention. At the 0th and8th week fasting blood sample and anthropometric measures were collected.

Results: the body weight, waistto hip ratio, fasting glucose levels, LDL decreased significantly (p<0/05), while HDL.C and Sirtuin-1 increased significantly (p<0/05). On the other hand, the 8 week aerobic exercises and the consumption of resveratrol supplement led to a significant increase in Sirtuin-1and a significant decrease in fasting glucose levels and LDL cholesterol. There were no significant changes in other variables in this group (p>0.05). In both of the experimental groups C reactive- protein did not have a significant change (p>0.05).

Conclusion: these findings show that probably, the consumption of resveratrol supplement has no effect on the Sirtuin-1expression but eight weeks aerobic exercises increments the expression and activity of this enzyme.

Key words: *aerobic exercises, Rezvin (resveratrol), Sirtuin-1, type 2 diabetes,*

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