diet and disease relationships. Biomarkers of dietary exposure should be valid, reproducible, able to detect changes in intake over time and be suitable for the general population. Yet many of the dietary biomarkers reviewed appeared inadequate at meeting all of the mentioned criteria. There are multiple factors that warrant investigation before many of these biomarkers can be more widely utilized in nutrition and health research. Genetics, age, type of specimen, time of year, and confounding dietary sources play a pivotal role in the feasibility and validity of dietary biomarkers. Future research should be directed at refining existing biomarkers by accounting for confounding factors, establishing new indicators of specific food intake and developing techniques that are cost-effective, noninvasive, rapid and accurate measures of nutritional status.

D-22

PITUITARY BIOMARKERS IN HEALTH AND IN PITUITARY DISEASES

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The pituitary gland produces and secretes hormones those play fundamental roles in regulating endocrine function. The pituitary gland has two lobes; an anterior and a posterior lobe. Adrenocorticotropic hormone (ACTH), growth hormone (GH), thyroid-stimulating hormone (TSH), luteinizing hormone (LH), folliculo-stimulating hormone (FSH) and prolactin (PRL) are secreted from the anterior lobe. Posterior pituitary releases antidiuretic hormone (ADH)/vasopressin and oxytocin, which are synthesized by the neurosecretory cells in the hypothalamus and stored in the posterior pituitary. ACTH stimulates synthesis and secretion of glucocorticoids, mineralocorticoids and androgens from the adrenal cortex. The most important secretagogue of ACTH is corticotropin releasing hormone (CRH). Physical, emotional and chemical stresses stimulate ACTH secretion. ACTH has a pulsatile secretion pattern along with the CRH. It has a diurnal rhythm. Hypersecretion of ACTH results in Cushing Disorder and hyposecretion results in secondary adrenocortical insufficiency. GH synthesized and secreted by the somatotroph cells within the anterior lobe. Growth hormone releasing hormone (GHRH), ghrelin and somatostatin influence the secretion of GH. Primary function of GH is the promotion of linear growth. Growth-promoting effects are mediated mostly by insulin-like growth factor 1 (IGF-1), but it has direct effects too. GH deficiency results in dwarfism in children. In the adults GH deficiency results in several metabolic disturbances and osteoporosis. On the other hand gigantism or acromegaly develops with GH excess. The hypothalamic control of PRL secretion is mainly inhibitory. Dopamine is the major inhibitory factor. The major function of PRL is stimulating lactation in the postpartum period. Hyperprolactinemia in adults results in hypogonadism. Absence of the lactation is the major consequence of hyperprolactinemia. TSH secretions is controlled by TRH (stimulates) and somatostatin (inhibits). Thyroid hormones control secretion by negative feedback. TSH deficiency causes central hypothryoidism and with TSH excess inappropriate TSH secretion results in thyrotoxicosis. LH and FSH regulates sex steroid secretion and gametogenesis. Deficiency of gonadotropins results in hypogonadism in both sexes. Excess gonadotropin secretion causes hypogonadism, ovarian hyperstimulation or testicular enlargement. Pituitary hormones should be evaluated along with the target gland hormones. Pulsatile secretion and short plasma half lives deserve attention while interpreting the results. For the evaluation of pituitary disorders dynamic tests are needed along with the basal hormone levels.

D-23

LABORATORY POINT OF VIEW IN PITUITARY / ADRENAL AND GONADAL DISEASES

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Hormones are organic compounds secreted into the bloodstream by specific glands, which modulate the functions of the tissues and specific organs that they reach via blood and act in very low quantities. It is derived from the Latin word 'hormaein', to stimulate, to act. The tissues in which they function are called target tissue. Some hormones may not be released from a specific gland or may have local effects instead of secretion into blood. Endocrinology is the study of medicine that relates to the endocrine system. The production and release of hormones into the blood are regulated by hierarchical control mechanisms. The majority of the hormones are released into the bloodstream by the effect of control mechanisms moving from top to bottom. The top step of these control mechanisms is the hypothalamus located at the base of the brain. With different neural stimuli reaching the cholestatic region, this region leads to the release of very small amounts of specific hormones, which we call releasing-releasing (sometimes slowing-inhibiting) factors. These hormones reach the anterior lobe of the 'hypophysis', a small endocrine gland located in the bone space called Sella Turcica that placed in the middle region of the brain via nerve fibers. Each secretory factor secreted by the hypothalamus leads to the release of a specific hormone from the anterior pituitary gland. The hormones released from the pituitary gland reach to the target tissues and glands via bloodstream and perform their specific functions. These functions are often as to stimulate the target gland for the production and release of its own hormones. Some hormones are not subject to this hierarchical system or are very little dependent. There are different stimulating and inhibiting mechanisms that regulate the synthesis and release of these hormones such as insulin, epinephrine, and glucagon.

D-24

NOT A MYTH, BUT A DISEASE WE IGNORE; PORPHYRIAS

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Porphyrias are a group of rare metabolic disorders characterized by the lack of enzymes involved in the synthesis of 'haem' and the excessive accumulation of haem precursors before the defective step. Diagnosis is usually delayed or it is likely that porphyria is often not considered at all as a cause of the patient’s symptoms and relevant patients may thus not be tested for these disorders. The presence of different clinical types, and the emergence and variation of symptoms relevant to many different medical specialties complicate the accurate diagnosis. The fact that the number of patients diagnosed with porphyria in our country is very low compared to Europe is probably due to the limited awareness of the physicians about the disorder and the lack of sufficient specialized laboratories to diagnose porphyria. Currently, prevention of acute attacks is possible with preventive measures and treatments if the patient is accurately diagnosed, but unfortunately patient’s quality of life is very low because of the lack of accurate diagnosis in most cases. It is of great importance that the patients are diagnosed so that the screening of relatives and genetic counseling can be carried out especially in consanguineous marriages. European Porphyria Initiative (EPI), founded in 2001 to improve the quality of diagnosis and treatment of porphyria patients in Europe, has been active as European Porphyria Network (EPNET) since 2007. The organization, which has succeeded in creating an effective network of specialized porphyria centers within the European Union, includes porphyria centers of 21 countries working to develop a current consensus-based approach for management for the disease, patients and their families. Many European countries with much smaller population than Turkey, have reported EPNET a much higher number of patients diagnosed with porphyrias than Turkey which points out how our health system
ignores the disorder. If a "selective screening" including patients with suspected symptoms, which is the recommended method for detection of rare diseases is carried out, data on the prevalence of the disorder in Turkey can be obtained and porphyria patients may receive the accurate diagnosis/treatment without wasting time and proper living conditions, taking necessary precautions may be assured.

D-25
ARTIFICIAL INTELLIGENCE BASED APPROACHES IN MEDICAL LABORATORIES: UNIVERSITY OF HEALTH SCIENCES TEPECK TRAINING & RESEARCH HOSPITAL EXPERIENCES

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Artificial Intelligence (AI) is defined as software systems that improve the current processes with more efficient use of information and reduced costs by supporting more accurate decisions, or taking these decisions directly. By using AI technology, systems can be created that can perform certain human behaviors (such as device use) and mimic the process of thinking on an area of expertise (such as medical diagnosis). Despite known since 1960's, significant increase in AI use has been made possible by the more economical and powerful computer systems. It is expected that one of the main areas where AI technology is widely used is medicine. The processes that AIs are often be used in medicine are: optimization of laboratory and radiological analyzes, medical diagnosis, personalized treatment, treatment monitoring, robotic surgery, digital consultation, drug design, medical data management. It is possible to increase the quality in the medical laboratories with the improvements in phlebotomy unit, which is one of the important component of the preanalytical process, and which plays a significant role in laboratory errors. The contributions of AI to the medical laboratory organization will be assessed using AI technology in the phlebotomy unit visited by an average of 1000 patients per day. Our web-based test database that inform any knowledge regarding the test requested fast and effortlessly to clinicians, laboratory staff, health personnel and patients upon online and / or mobile environments will also presented. Finally, "Predictive Quality", a new approach to internal quality control evaluation developed by faculty members from 9 Eylül University, Department of Computer Engineering and Medicine Faculty Department of Medical Biochemistry and tested in our laboratory will be discussed. In summary, it is planned to increase awareness of AI in medicine by sharing our experience on the AI that is expected to improve quality and productivity in health.

D-26
CLINICAL APPROACH TO INBORN ERRORS OF METABOLISM

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Disorders of inborn errors of metabolism are a group of rare diseases caused by defects of coding genes of enzymes, transport proteins or structural proteins of the body. Because of the high rate of consanguinity and the high rate of the reproductivity, metabolic disorders are more common in Turkey than the rest of the world. According to their pathogenesis, metabolic disorders can be divided into three diagnostic groups; intoxication type diseases, disorders related with energy metabolism and disorders related with complex molecules. Generally, these diseases can be occurred in four different clinical presentations: 1) Early symptoms in the antenatal and neonatal period (hydrops fetalis, sepsis etc), 2) Later onset acute/ recurrent attacks of symptoms (coma, ataxia, vomiting, acidosis, cardiac, renal, liver or other organ failure), 3) Chronic and progressive neurological symptoms (developmental delay, mental retardation, seizures, psychiatric symptoms), 4) Specific and permanent organ/system presentations (such as cerebral, ocular, renal, cardiac, hepatic signs). After the diagnosis of metabolic disease, disease-specific treatment is used to the patients.

D-27
BIOCHEMICAL APPROACH TO INHERITED METABOLIC DISEASE- LABORATORY DIAGNOSIS

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There is a great spectrum of inherited metabolic diseases due to defects in enzymes/proteins related to about all biochemical pathways. The diagnosis of these disorders is performed at three stages, measurement of the metabolites, measurement of the activity of the responsible enzyme and the analysis of the specific mutation. Measurement of the metabolites and enzyme activities is also used for screening and monitoring of these diseases. Tandem Mass Spectrometry (MS/MS) is being widely used for newborn screening of inherited metabolic diseases. Mass spectrometry is an analytical technique in which molecules or fragments are defined and measured quantitatively according to mass-charge ratio. It is possible to screen many metabolic diseases including primary aminoacidemias, urea cycle disorders, organic acidemias and fatty acid oxidation disorders by analysis of dried blood spots by MS/MS. The reference method for quantitative analysis of amino acids consists of ion exchange liquid chromatographic separation followed by photometric measurement of ninhydrin reaction and is performed by amino acid analyzers. Recently, Liquid Chromatography Tandem Mass Spectrometry (LC-MS/MS) is being more widely preferred for amino acid analysis due to its speed. Urinary organic acids are analysed by Gas Chromatography Mass Spectrometry (GC-MS). In addition to these basic analyses, many other metabolites are analysed using mass spectrometric techniques eg. fatty acids by GC-MS; purins and pyrimidines, bile acids and steroids by LC-MS/MS. In metabolism laboratories, high performance liquid chromatography is used for the analysis of oxalate and strate, glycosylated proteins, electrophoresis is used for separation of mucopolysaccharides and thin layer chromatography is used for separation of oligosaccharides. In some storage disease, the abnormal metabolites cannot be detected, thus direct measurement of the enzyme activity is performed in leucocyte or fibroblast homogenates or in serum or plasma. In recent years LC-MS/MS is being applied for measurement of lysosomal enzymes in dried blood spots. In metabolism laboratories; spectrophotometric and spectrofluorimetric methods are also still valid for measurement of enzyme activity eg. Biotinidase and for measurement of some metabolites eg. sialic acid, mucopolysaccharides. The diagnosis of inherited metabolic disease requires the use of numerous different techniques for the analysis of molecules representing different biochemical pathways.

D-28
BIOCHEMICAL APPROACH TO INBORN ERRORS OF METABOLISM

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Inherited metabolic disorders are a growing number of disorders with increasing diagnostic methods usually seen in newborns or early childhood. These disorders can lead to permanent physical and mental retardation, coma and death if not treated. Early diagnosis is important in terms of success in treatment and prevention of permanent sequelae. Some of the diseases can be diagnose with newborn screening tests, while others occur with clinical symptoms. These diseases can cause life-threatening conditions in the acute phase. Emergency tests are of great importance for an undiagnosed patient, while patients with known anomalies are sometimes presenting with acute episodes. In such cases, patients must be evaluate urgently. The most common clinical presentation of inherited metabolic diseases are nonspecific conditions such as cataleptic state due to acute decompensation, energy deficiency, and acidosis. Emergency tests such as whole blood count, electrolytes, blood sugar, calcium, blood gases, uric acid, PT, liver function tests, ammonia, lactic acid, pyruvic acid, creatine kinase, pH are used for evaluation and treatment of this