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 TEPECIK 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 for 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 striate, glycosilated proteins, electrophoresis is used for separation of mucopolysaccharidases and thin layer chromatography is used for separation of oligosaccharidases. 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 spectrophotometric methods are also still valid for measurement of enzyme activity eg. Biotinidase and for measurement of some metabolites eg. sialic acid, mucopolysaccharidases. 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 catabolic 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
condition. In addition, many analyses such as odor, reductant substance, ketoacids, sulfite test and phenylpyruvic acid test are also used. In addition to these tests useful in the acute phase, many metabolites such as amino acids, fatty acids and organic acids can also be measured using advanced technological chromatographic techniques to guide the diagnosis and treatment of the disease. As a result, early diagnosis and emergency laboratory evaluation is the most important step for success of treatment. A missed diagnosis can lead to lifelong sequel and even death.

D-29
BIOMARKERS IN LYSOSOMAL STORAGE DISEASE: LABORATORY APPROACH
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Lysosomal storage diseases which are related to deficiency of specific lysosomal hydrolases resulted to clinical aspects due to accumulation of substrates in different tissues. Since Dried Blood Spot (DBS) is non-invasive, low-cost, easy transportable, acceptable enzyme stability compared to leucocyte and/or fibroblast culture, it’s recommended as a first screening test. As enzyme replacement therapies are available currently, early diagnosis of these diseases is crucial nowadays. The gold standard for diagnosis is determination of enzyme activity in DBS and/or plasma and/or leucocyte samples. Disease diagnosis is verified by determination of genetic mutation in gene of enzyme protein which is specific for LSD. However, a variety of problems such as low accuracy of enzyme activity methods, unknown genetic mutations, high ratio of false positive diagnosis due to methods, complicate the correct diagnosis of these patients. Therefore, clinicians need new biomarkers other than enzyme activity to diagnose and monitor of enzyme replacement therapy of the patients. Recently two types biomarker have been suggested for LSD, 1) Primer biomarkers which are metabolites accumulated in tissue due to enzyme deficiency, found in plasma and/or urine, e.g. glycosaminoglycan in urine of patients with mucopolysaccharidosis, tetrasccharide in urine of patients with Pompe disease. 2) Secondary biomarkers which are non-specific, increase in serum/urine resulted from damaging of other tissues due to disease, e.g. biomarkers of liver damage and renal damage. Some biomarkers in this group are partially specific to disease, e.g. chitotriosidase which is a macrophage activation marker, increases in blood of patients with Gaucher disease, Niemann Pick disease. Currently, LAMP-1, LAMP-2, some interleukins, saposins and cathepsins as further biomarkers are focus of investigations.

D-30
BIOMARKERS FOR LYSOSOMAL STORAGE DISORDERS: CLINICAL APPROACH
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Lysosomal storage disorders (LSD) are group of diseases with metabolic defects associated primarily with a disruption in the catabolism and/or transport of by-products of cellular turnover, coupled with the secondary consequences of the accumulation of incompletely metabolized substrates within particular cell types. Initially, the individual disorders were grouped according to the chemical composition of the storage material, e.g. sphingolipidosis (Gaucher, Fabry, Niemann-Pick A/B/C, Metachromatic leukodystrophy, Krabbe disease, Tay-Sachs/Sandhoff disease, GM1-gangliosidosis), mucopolysaccharidoses (Hurler/Scheie, Hunter, Sanfilippo, Morquio, Maroteaux-Lamy, Sly, Nataowitz) oligosaccharidoses (Mannosidosis, Sialidosis, Fucosidosis, Aspartylglucosaminuria ) etc. More recently, these disorders have been clustered according to their biochemical or molecular basis. To date, the LSDs encompass at least 250 different clinical entities. LSDs are pernicious, multi-systemic and under diagnosed disorders, frequently with a (sub) clinical onset at pediatric age. Their phenotype is heterogeneous in age of onset, rate of progression and involved organs. Several clinical manifestations, such as hepatosplenomegaly, coarse facial features and skeletal dysplasia, can serve as an important clue for LSD. On presentation, especially in a young child, the diagnosis can be missed, particularly when the family history is uninformative. Therefore, identification of the biomarkers that can serve as a surrogate for or indicator of disease severity, in terms of either overall disease burden or involvement of a particular organ or system is very important. Diagnostic confirmation necessitates biochemical and/or molecular genetic testing. Ideally biomarkers should be easily and cheaply measurable in readily obtained samples (urine/blood) and moreover, their concentration or activity should be found to be greatly elevated in disease states, without overlap in values between affected and healthy subjects, and should change rapidly in response to specific treatment outcomes that are clinically meaningful. The main known markers for LSD are chitotriosidase-CLC18-PARC-ACE-TRAP(Gaucher), globotriaosylceramide (lysoGb3)-uromodulin (Fabry) and urinary Glc4-plasma Hex4 (Pompe).

D-31
GENETIC APPROACHES FOR INHERITED METABOLIC DISEASES
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Genomic or mitochondrial abnormalities cause congenital disorders and one of the major group of these disorders is inherited metabolic diseases. Metabolic pathways are important for continuity for an organism and particular abnormalities on the pathway affect function of specific proteins and/or enzymes. Inherited metabolic disorders are monogenic disorders which are mainly autosomal recessive manner. Autosomal dominant and X linked patterns affect metabolic pathways less frequently. Small DNA changes can affect inborn errors of metabolism by anomalies of nucleotides which are important for their functionality. For research and/or routine clinical diagnosis, mainly molecular techniques such as Sanger and next generation sequencing are used recently. Molecular genetic analyses and their techniques are highly important issue for diagnosis because it is necessary for genetic counselling and evaluating appropriate treatment opportunities.

D-32
CULTURE OF PRODUCTION
Yasin Yolcu
Rel Assay Diagnostics

Our company founded in 1993 based on all branch of medical sector, but we started the transition for laboratory service because this field was süit for our educate. As we manufacturer company, our thinking based on provide our national culture of production, high technology, sustainable development and inheritance by future generations. We observed that there were gap of scientific and industry and there were not any meeting platform. We observed the Prof. Dr. Özcan EREL’s works on diagnostics field and negotiated the work for industry. Prof. EREL decided that agreed with us for benefit of our national. This was initial model and New step for our country because there were not national precedent on international so many of our decisions and behaviors were first for our country. Our platform of inventors expanded with Doç. Dr. Şahabettin GEKİK in time. We observed many unknown before the production so we solved step by step. We attained international fuar and congress as Turkish company. We decided that take the quality certificates such as KFDA however scientists used the our product so we have many international articles by the way the value of our trade mark increased as rapidly. This event supported the our national high technology image for international area. Value of the