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 Eylul 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 aminoacidasides, urea cycle disorders, organic acidasides 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 olosasacharides. 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 the 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 catacolic 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