Full blood count, biochemical analysis, thyroid function, and pubertal hormone tests were found to be normal in terms of tall stature. Serum levels of IGF-1 and IGFBP-3 ranged from 0 to +1 SDS. Echocardiography revealed mitral valve prolapse. The eye examination was normal in terms of lens subluxation.

In this report, Marfan syndrome with tall stature and transverse striae of the back was presented. Early diagnosis and appropriate treatment will prevent the development of complications.

(P-38)

Diagnostic Algoritm in Two Different Cases with Subclinical Endocrinologic Problems

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Incomplete form of Di George syndrome (DGS) which is characterized by hypoparathyroidism, thymic aplasia, facial dysmorphism, and cardiovascular anomalies may present with subclinical endocrine problems and may result in delay in diagnosis.

Case 1: A 2-year-old girl born to unrelated healthy parents was consulted due to mild hypocalcemia before angiographic evaluation for tetralogy of Fallot. Anthropometric evaluation was appropriate for her age. She had perioral cyanosis, mild hypertelorism, high palate, and minor anomaly in her toe. She had borderline hypocalcemia, low parathormone, and high TSH levels in biochemical and hormonal evaluation. Thymus was absent in her chest X-ray. CD3, CD4, CD8, CD19 were low, total complement level, quantitative immunoglobulin levels, and in vitro lymphocyte transformation tests were normal. **Case 2:** A 31-day-old female patient born to unrelated healthy parents was consulted due to high TSH levels. Anthropometric evaluation was appropriate for her age. She had clubfoot, low-set ears, micrognathia, and high palate. In laboratory evaluation, hypocalcemia and high TSH level were determined. Ostium secundum ASD and bilateral hydronephrosis were observed in echocardiography and renal ultrasonography. Thymus gland was present in chest X-ray.

DOUBLE FISH analysis was performed. Case 1: Heterozygote 22q11 mutation was determined and the patient was diagnosed as having incomplete DGS. Case 2: Homozygote 22q11 deletion was determined and the patient was diagnosed as having DGS.

It's important to perform DOUBLE FISH analysis in cases with subclinical endocrine problems if incomplete DGS is suspected. Thus, major problems (such as graft versus host disease due to transfusion), which may patient face in the future, might be prevented.

(P-39)

A Novel *De Novo* Missense Mutation in HNF4A Resulting in Sulfonylurea-Responsive MODY

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Maturity-onset diabetes of the young (MODY) is a monogenic form of diabetes with autosomal dominant inheritance and usually develops before 25 years. Heterozygous inactivating hepatocyte nuclear factor 4A (HNF4A) mutation is a rare subtype of MODY.

A 14-year-old girl was admitted to our clinic due to fatigue and polyuria, and hyperglycemia was detected afterward. She was born full term with birth weight of 5500 g (4.9 SD score) and had no postnatal hypoglycemia. Her parents were not relatives and family history revealed no diabetes. Physical examination revealed a height of 163 cm (SD score 0.29), weight 64.7 kg (SD score 1.2), and body mass index of 24 kg/m^2 (SD score 1.2). Neither acanthosis nigricans nor striae were found. Laboratory analyses showed C-peptide 1.66 ng/mL (normal, 0.9-7.1 ng/mL), glycated hemoglobin (HbA1c) 8.8%, normal lipid profile, and negative autoantibodies regarding diabetes. Urine analysis showed 2+ glycosuria and no ketosis. We started only insulin glargine (0.2 unit/kg/day) with most probable diagnosis of MODY. Normal glycemia was improved and no hypoglycemia was seen with this treatment. HbA1c was decreased to 6.3%.

Genetic analysis revealed a *de novo* p.C93Y (c.278G>A) heterozygous novel change in the HNF4A. Insulin treatment was stopped and low-dose sulfonylurea (5.0 mg/day) initiated when the diagnosis of MODY 1 was proved. After five months of the treatment onset, fasting glucose was 111 mg/dL, insulin 11 IU/mL, C-peptide 2.2 ng/mL, and HbA1c was 5.8%.

Genetic testing should be considered to establish an accurate diagnosis and provide an opinion to determine the appropriate type of treatment.