NURSING PRESENTATIONS

Sp 18

Haematological emergencies and the early warning score tool for nurses

Dana Parness

This talk will focus on major hematological emergencies, and the nurse's role in recognizing the red flags, responding acutely and treating, while educating the next generation

I will discuss three main hematological emergencies using real life clinical cases, to review the relevant classification scores, risk factors, and treatment approach- enabling better understanding of the nurse's role in early diagnosis and treatment

TLS-tumor lysis syndrome- Recognizing patients at risk for TLS, and monitoring them for early signs of TLS, while applying prevention strategies, such as Allopurinol and hydration for high-risk patients. Using the Cairo Bishop classification score for definite diagnosis, using clinical and laboratory data. We will further discuss, how to best monitor and treat patients with established TLS, as recommended by the latest guidelines, through the different clinical cases presented.

SVCS- superior vena cava syndrome

We will discuss the myriad of symptoms and signs pointing to SVC syndrome, the different risk factors, and etiologies, as well as the differential diagnosis and approach to establishing a definite diagnosis. As abovementioned, using clinical cases we will discuss the updated approach to diagnosis and treatment, focusing on the nurse's crucial role in prevention and early response and management

Neutropenia fever-sepsis

Viewing this crucial subject in treating hematological patients, we will discuss in depth the definitions of neutropenia, and the different scores used to delineate sepsis, septic shock and severe shock. We will then continue to case studies, exploring the approach, to the febrile neutropenic patient- starting from the basic management, eg hemodynamic monitoring, fluid resuscitation, laboratory workup. Thereafter, we will continue to practice early signs of sepsis and grades of shock, then covering in brief the diagnostic possibilities and various treatment regimens available, by clinical, laboratory and imaging studies, stressing the importance of thorough physical exam, blood cultures, early initiation of broad-spectrum antibiotics, and adequate fluid balance. To summarize this subject, we will discuss different strategies for prevention of sepsis, and the crucial role of the nurse team in diagnosing, treating and preventing complications

We will close the meeting, with a summary of the abovementioned subjects, emphasizing the importance of the nurse's role in every patient case- whether by frequent monitoring, attention to subtle changes or by creating close and trusting relationships with the patients and their caregivers, all of which enable early recognition and intervention, crucial to the lives of our patients.

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Sp 19

How to COVID pandemic changed clinical practice

Medine Yılmaz

The corona virus disease (COVID) 19 pandemic has affected the entire health system and the delivery of health services. This influence has brought about the change in clinical practices. Nurses are essential in the fight against the patient care, COVID 19. The management of COVID 19 has shown some differences according to the countries' health systems and health manpower. In this context, this presentation talk will focus on the reflections of the COVID 19 pandemic on clinical practices in Turkey.

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PEDIATRIC PRESENTATIONS

Sp 09

Congenital neutropenias: Türkish Registry

Deniz Yılmaz Karapınar

Severe congenital neutropenia is a rare disease, and autosomal dominantly inherited. ELANE mutation is the most frequently observed genetic defect in the registries from North America and Western Europe. However, in eastern countries where consanguineous marriages are common, autosomal recessive forms might be more frequent.

Two hundred and sixteen patients with severe congenital neutropenia from 28 different pediatric centers in Turkey were registered. Patients inclusion and exclusion strategies apre shown in Figure-1.

The most frequently observed mutation was HAX1 mutation (n = 78, 36.1%). A heterozygous ELANE mutation was detected in 29 patients (13.4%) in our cohort. Biallelic mutations of G6PC3 (n = 9, 4.3%), CSF3R (n = 6, 2.9%), and JAGN1 (n = 2, 1%) were also observed (Table 1). Eighty seven percent of HAX1 mutations were detected in the same point of p.

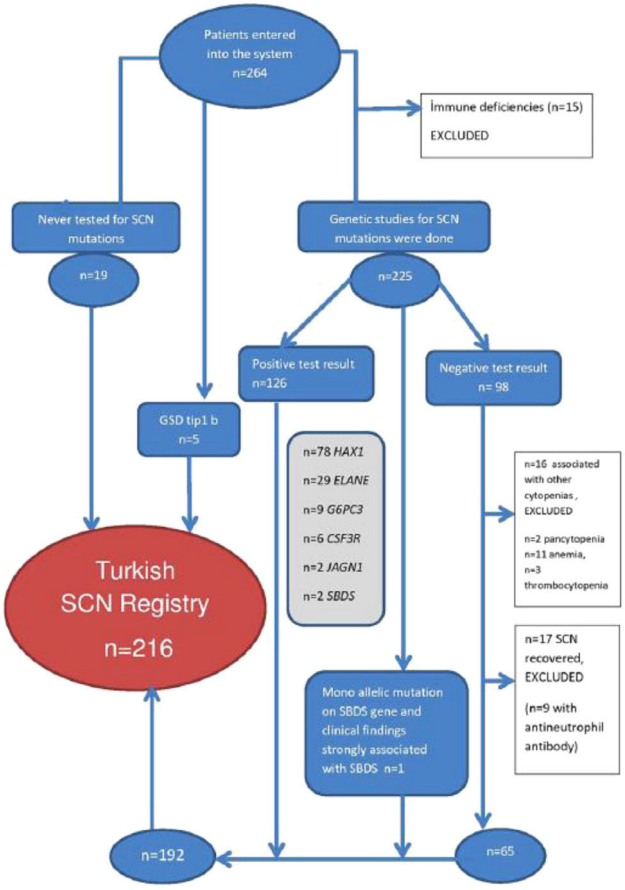
Table 1 – Congenital Neutropenia mutations and their frequencies in the Turkish Severe Congenital Neutropenia Registry

Mutation analyses	n(%)
HAX1 (+)	78(36.1)
ELANE (+)	29(13.4)
G6PC3 (+)	9(4.2)
CSF3R (+)	6(2.8)
JAGN1 (+)	2(1)
ELANE-/HAX1-	23(10.6)
ELANE-/HAX1-/G6PC3-	20(9.3)
ELANE-/HAX1-/G6PC3-/JAGN1-/CSF3R-	22(10.2)
GSDtype 1b	5(2.3)
SBDS	3(1.4)
No genetic testing performed	19 (8.8)

W44X. There were 6 patients who had a HAX1 mutation other than c.130-131 pW44X point mutation. Four had novel mutations. The novel mutations were detected in 7 ELANE patients. Interestingly 2 out of 4 patients with CSF3R mutation and 4 out of 9 patients with G6PC3 had a novel mutation.

Granulocyte colony-stimulating factor treatment was given to 174 patients (80.6%). Two patients died with infectious complications, and five patients developed myelodysplastic syndrome/acute myeloblastic leukemia (Table 2). The mean (± mean standard error) follow-up period was 129.7 ± 76.3 months, and overall survival was 96.8% (CI, 94.4-99.1%) at the age of 15 years.

In Turkey, mutation analysis should be started with HAX1, and if this is negative, ELANE and G6PC3 should be checked. Because of the very high percentage of consanguineous marriage, rare mutations should be tested in patients with a negative mutation screen.



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Sp 10

Hereditary macrothrombocyte disorders

Zühre Kaya

Hereditary macrothrombocyte disorders as known inherited macrothrombocytopenia (IMTP) are gaining greater recognition through advanced genetic and molecular studies. It is a heterogenous group of rare bleeding disorders characterized by abnormally giant platelets, thrombocytopenia, mild to moderate bleeding phenotypes, and/or positive family history and/or syndromic findings. The main characteristic features of macrothrombocytopenia are reduced platelet count (<150,000/μL) and significantly enlarged platelets (mean platelet volume (MPV) >12fL). Up to now, more than 30 genes linked to IMTP have been identified in nearly half of the patients with syndromic and non-syndromic IMTP. All inheritance patterns, including autosomal dominant, recessive, and sex-linked, have been described; however, nearly 50% of affected patients have unidentified genetic mutations or molecular abnormalities. The early and late stages of megakaryopoiesis and subsequent proplatelet formation and functional platelet are regulated by a large number of genes. Defects in these genes result in the dysfunction of several steps in megakaryopoiesis, proplatelet formation, and mature platelet. Many patients with non-syndromic IMTP are either asymptomatic or have minor bleeding manifestations and, detected incidentally with platelet count and morphology evaluation; however, patients with syndromic IMTP may be early diagnosed by specific clinical findings in addition to macrothrombocytopenia. Physicians should suspect IMTP if the following clinical and laboratory findings are present:

a Hearing loss, cataracts, impaired renal function, elevated hepatic enzymes and Döhle-like bodies in neutrophil for non-muscle myosin heavy chain (MYH)-9 related disease,
b Splenomegaly, bone marrow fibrosis, pale platelet and elevated serum levels of vitamin B12 for gray platelet syndrome,
c Cardiac abnormalities, dysmorphic face, digital abnormalities and mental retardation for Paris Trousseau thrombocytopenia/Jacobsen syndrome,
d Craniofacial defects, cardiac abnormalities, mental retardation, hypotonia, thymic aplasia, immune deficiency for DiGeorge syndrome/Velocardiofacial syndrome,
e Xanthomas, premature atherosclerosis, arthritis, hemolytic anemia with stomatocytes, elevated plasma phytosterols for sitosterolemia.

No definitive guidelines are available for managing asymptomatic or mildly symptomatic patients with IMTP; however, platelet transfusion, antifibrinolytic therapy, and recombinant factor VIIa are usually recommended for those with severe bleeding manifestations or prior to surgery. Recently, it has been reported that thrombopoietin-receptor agonists can be used in some patients with MYH9 related disorders. The International Society of Thrombosis and Haemostasis' scientific subcommittee on platelet physiology has published guidelines for diagnosing IMTP. In this approach, personal and family history, and physical examination are the key, followed by i) investigation of platelet count and morphology, ii) evaluation of platelet function by light transmission aggregometry, and iii) a panel of tests to assess granule contents and platelet surface markers by electron microscopy and flow cytometry studies. Further, next-generation sequencing has greatly expanded the molecular repertoire of IMTP, thus enabling the identification of new disorders.