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Thromboembolic Diseases in Rare Disorders and its Diagnosis

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Description

Thromboembolic diseases, characterized by the formation of blood clots that obstruct blood flow, represent a significant health concern worldwide. While commonly associated with conditions like deep vein thrombosis or pulmonary embolism, these disorders can also manifest in rare diseases, presenting unique challenges in diagnosis, management, and treatment. Thromboembolic diseases arise from the disruption of normal blood flow due to the formation of blood clots in blood vessels [1]. These clots can travel through the bloodstream and lodge themselves in vital organs, leading to severe complications such as stroke, heart attack, or organ damage. The incidence of thromboembolic events is relatively high in the general population, often linked to factors like prolonged immobility, surgery, or underlying cardiovascular conditions [2]. In rare disorders, the risk of thromboembolic events can be heightened due to specific disease-related factors. Many rare diseases are characterized by alterations in blood composition, vessel structure, or clotting mechanisms, increasing the likelihood of clot formation. Disorders such as Antiphospholipid Syndrome (APS), paroxysmal nocturnal hemoglobinuria or certain genetic coagulation disorders exemplify conditions where thromboembolic events are prevalent yet often overlooked due to their rarity [3].

Antiphospholipid syndrome

APS, an autoimmune disorder where the immune system mistakenly targets phospholipids, significantly increases the risk of thrombosis. Patients with APS may experience recurrent miscarriages, arterial or venous thrombosis, and other pregnancy-related complications [4-6]. The management of APS involves anticoagulant therapy to prevent clot formation and reduce the risk of thromboembolic events, emphasizing the importance of early diagnosis and treatment. PNH is a rare acquired disorder of the blood characterized by the abnormal breakdown of red blood cells due to increased sensitivity to complement, a part of the immune system. Patients with PNH are at heightened risk of thromboembolism, particularly in the veins [7]. Treatment involves complement inhibitors and anticoagulation therapy to manage both the hemolytic symptoms and reduce thrombotic risk. Genetic disorders affecting blood clotting factors, such as Factor V Leiden mutation mutation or deficiencies in antithrombin, protein C, or protein S, predispose individuals to thromboembolic events from a young age. These conditions often necessitate lifelong anticoagulation therapy and careful management to mitigate the risk of clot formation in arteries and veins[8].

Diagnosis

Diagnosing thromboembolic events in the context of rare disorders can be challenging due to their diverse clinical presentations and the overlap with other disease symptoms. Furthermore, the management of thrombotic risk in rare diseases requires a tailored approach that considers both the underlying disorder and individual patient factors. Clinicians face the dual challenge of balancing the need for anticoagulation with the risk of bleeding complications, requiring regular monitoring and adjustment of treatment strategies. Advancements in understanding the pathophysiology of thromboembolic diseases in rare disorders have led to targeted therapies and improved outcomes. Research efforts focus on identifying novel biomarkers, exploring genetic predispositions, and developing personalized treatment strategies to minimize the burden of thrombotic events in vulnerable patient populations [9,10]. Collaborative initiatives between researchers, clinicians, and patient advocacy groups are pivotal in advancing knowledge and improving clinical care for individuals affected by these challenging conditions. Thromboembolic diseases in rare disorders represent a complex intersection of thrombotic risk and unique disease pathologies. While these conditions pose significant challenges in diagnosis and management, advances in medical research and therapeutic interventions offer hope for improved patient outcomes. By raising awareness, promoting early detection, and fostering interdisciplinary collaboration, healthcare professionals can better navigate the complexities of thromboembolic diseases in rare disorders, ultimately enhancing the quality of life for affected individuals. The recognition and proactive management of thromboembolic risks in rare disorders are critical in mitigating severe complications and improving patient outcomes. Continued research and clinical vigilance are essential in addressing these challenges and advancing care for individuals living with rare diseases susceptible to thromboembolic events.

1. Camona C, Wilkins KB, Drogos J, Sullivan JE, Dewald JPA, et al. (2018) Improving hand function of severely impaired chronichemiparetic stroke individuals using task-specific training with the rein-hand system: A case series. Front Neurol 9.
2. Yun GJ, Chun MH, Park JY, Kim BR (2011) The synergic effects of mirror therapy and neuromuscular electrical stimulation for hand function in stroke patients. Ann Rehabil Med 3: 316.
3. Wolbrecht ET, Rowe JB, Chan V, Ingemanson ML, Cramer SC (2018) Finger strength, individuation, and their interaction: Relationship to hand function and corticospinal tract injury after stroke. Clin Neuro physiol 129: 797-808.
4. Israely S, Leisman G, Carmeli E (2017) Improvement in arm and hand function after a stroke with task-oriented training. BMJ Case Rep 10: 1136-1143.
5. Martin GM (1965) Stroke rehabilitation. S D J Med18: 20-26.
6. Kim J, Yim J (2017) Effects of an exercise protocol for improving handgrip strength and walking speed on cognitive function in patients with chronic stroke. Med Sci Monit 23: 5402-5409.
7. Yeldan I, Huseyinsinoglu BE, Akinci B, Tarakci E, Baybas S (2015) The effects of very early mirror therapy on functional improvement of the upper extremity in acute stroke patients. J Phys Ther Sci 27: 3519-3524.
8. Luo Z, Zhou Y, He H, Lin S, Zhu R, et al. (2020) Synergistic effect of combined mirror therapy on upper extremity in patients with stroke: A systematic review and meta-analysis. Front Neurol 11: 5-9.
9. Gurbuz N, Afsar SI, Ayaş S, Nur S, Cosar S (2016) Effect of mirror therapy on upper extremity motor function in stroke patients: A randomized controlled trial. J Phys Ther Sci 28: 2501-2506.
10. Tosi G, Romano D, Maravita A (2018) Mirror box training in hemiplegic stroke patients affects body representation. Front Hum Neurosci 11.