The Hierarchical Escalation of Vascular Access in Nijmegen Breakage Syndrome. – Case Report

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Background

Nijmegen Breakage Syndrome (NBS) is a rare genetically determined disease presenting with microcephaly, mild growth delay, premature ovarian insufficiency, combined immunodeficiency and considerable increase in the likelihood of developing early-onset malignancies, particularly of haematological origin.

The disease is caused by a loss of nibrin (p95) function, which normally targets the MRN complex to DNA double-strand breaks and interacts with ATM kinase. Defective nibrin prevents the repair of radiation-induced DNA damage, explaining the marked cellular hypersensitivity to X-rays and other forms of ionizing radiation

While the global prevalence is estimated at approximately 1 in 100,000, it is notably higher in Eastern European and Slavic populations, reaching up to 2 in 100,000 in Belarus.

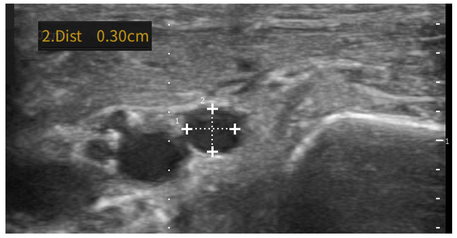
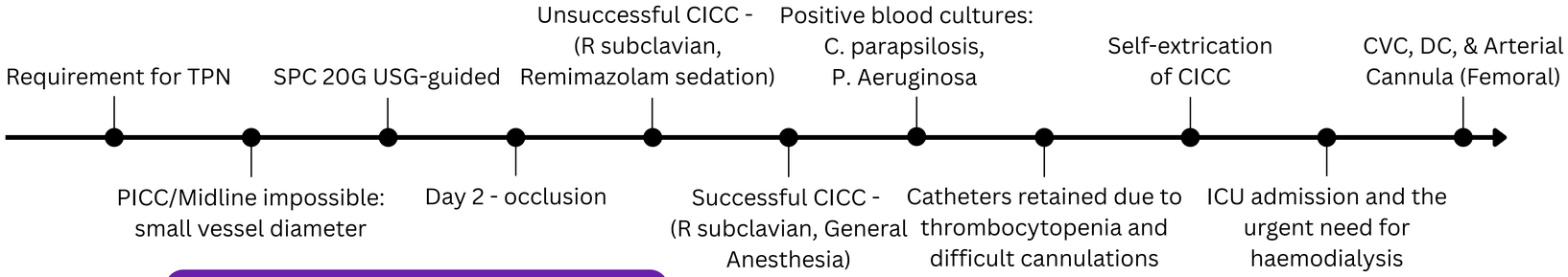


Fig. 1 Upper limb venous diameter ultrasound measurement



Case Report

An 18-year-old female patient, post-alloHSCT for Hodgkin lymphoma secondary to NBS, required the placement of an additional vascular access due to the necessity of concurrent continuous infusion and total parenteral nutrition (TPN).

Key Clinical Challenges:

- **Anatomical Barriers**: Small peripheral vessel diameter (<3mm) ruled out PICC/Midline options.
- **Hematological Risks**: Severe, non-correctable thrombocytopenia (NBS background) increased the risk of invasive procedures.
- **Patient Cooperation**: Neurological/behavioral factors necessitated General Anesthesia for central access.
- **Radiosensitivity**: US-guidance and IC-ECG tip verification adhering to ALARA

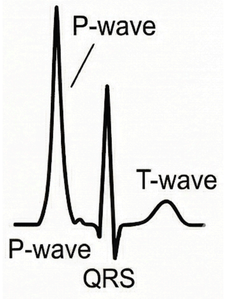


Fig. 2 Intra-atrial ECG guided CVC tip positioning

Conclusion

- **Universal Gold Standard**: A bespoke, hierarchical strategy integrating anatomical and behavioral factors should be the baseline for all vascular access patients.
- **Risk-Based Escalation**: Lack of cooperation (risk of self-extrication) and multi-organ failure (need for TPN) necessitate prompt escalation to secure central access.
- **Radiation-Free Safety**: Mandatory USG and IC-ECG tip verification are essential to maintain ALARA principles, especially in radiosensitive populations like NBS.

References

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