CASE REPORT

Management of a Patient with Noonan Syndrome and Von Willebrand Disease: A Case Report

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Abstract

Von Willebrand disease (VWD), the most prevalent inherited bleeding disorder, entails either a quantitative or qualitative defect in von Willebrand factor (VWF), which hinders platelet adhesion and aggregation and may subsequently lead to reduction in factor VIII levels. Noonan syndrome (NS) is a genetically heterogeneous condition that presents with distinctive craniofacial features like short stature, flat nasal bridge, widely placed eyes, webbed neck, congenital heart anomalies, and a predisposition to bleeding, which may be seen as coagulation factor deficiencies or platelet dysfunction. The combined presence of VWD and NS may act synergistically to increase the risk of hemorrhagic complications during dental procedures, particularly extraction, requiring meticulous interdisciplinary treatment planning and patient care. This case report emphasizes the indispensable need for a holistic and multimodal treatment approach for dental extractions and minimally invasive management of carious lesions in a pediatric patient with both VWD and NS, to ensure a safe and effective dental outcome.

Keywords: Bleeding disorder, Case report, Noonan syndrome, Pediatric patients, Von Willebrand disease.

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Introduction

Von Willebrand disease (VWD) is the most predominant inherited bleeding disorder, caused by either a quantitative or qualitative deficiency of von Willebrand factor (VWF), which helps in platelet adhesion and stabilization of factor VIII. This results in impaired primary hemostasis and, in some cases, reduced levels of factor VIII, increasing the risk of mucocutaneous and procedural bleeding.[1] Among the various subtypes, types IIA and IIM involve qualitative defects in VWF, leading to functional impairment despite normal or near-normal levels.[2]

Noonan syndrome (NS) is an autosomal dominant, genetically inherited, multisystem disorder with an incidence of 1 in 1,000 to 1 in 2,500 live births.[3,4] It is classified among the RASopathies due to mutations affecting the RAS/MAPK signaling pathway. NS is characterized by a distinct craniofacial phenotype: ptosis, down-slanting palpebral fissures, hypertelorism, low-set and posteriorly rotated ears, flat nasal bridge, short/webbed neck, and high-arched palate. Up to 80% of patients present with congenital heart defects, particularly pulmonary valve stenosis (PVS) and hypertrophic cardiomyopathy (HCM). NS is also associated with a predisposition to bleeding disorders, often due to platelet dysfunction or coexisting conditions such as VWD.[5]

The coexistence of VWD and NS presents peculiar challenges for dental management, particularly during invasive procedures such as extraction, due to increased risk of uncontrolled bleeding and cardiovascular complications. This case report highlights the importance of successful dental management of a 6-year-old male patient who was a known case of VWD type IIA or IIM and NS, which was genetically confirmed.

Case Description

A 6-year-old child presented to the pediatric and preventive dentistry department with a chief complaint of unpleasant smile due to multiple broken teeth in the upper front teeth region, noticed by his parents for the past 2 years. The patient did not complain of any pain or discomfort. Written informed consent was obtained from the parents prior to the examination and documentation.

A detailed medical history was taken from the parents and it was found that the patient had a significant medical history. He had a confirmed diagnosis of von Willebrand disorder type IIA or IIM, diagnosed 6 months back following prolonged bleeding from surgical site after surgery for undescended testes. He had a history of recurrent epistaxis, easy bruising, and prolonged bleeding from minor cuts. Previous management included Tranexa 500 mg, half a tablet.

Following that, the patient was investigated for bleeding disorder. The results showed complete blood count (CBC) normal, prothrombin time (PT) normal, activated partial thromboplastin time (aPTT) normal, factor XIII normal, but a reduced ristocetin cofactor activity for VWF to 26. Pediatric hematologist advised to avoid nonsteroidal anti-inflammatory drugs (NSAIDs), except for paracetamol, due to his bleeding disorder.

He also had a diagnosis of NS, genetically confirmed with a whole exome sequencing showing chr2:39285837C>T, c.322G>A, p.Glu108Lys, heterozygous, dominant. The patient's father reported

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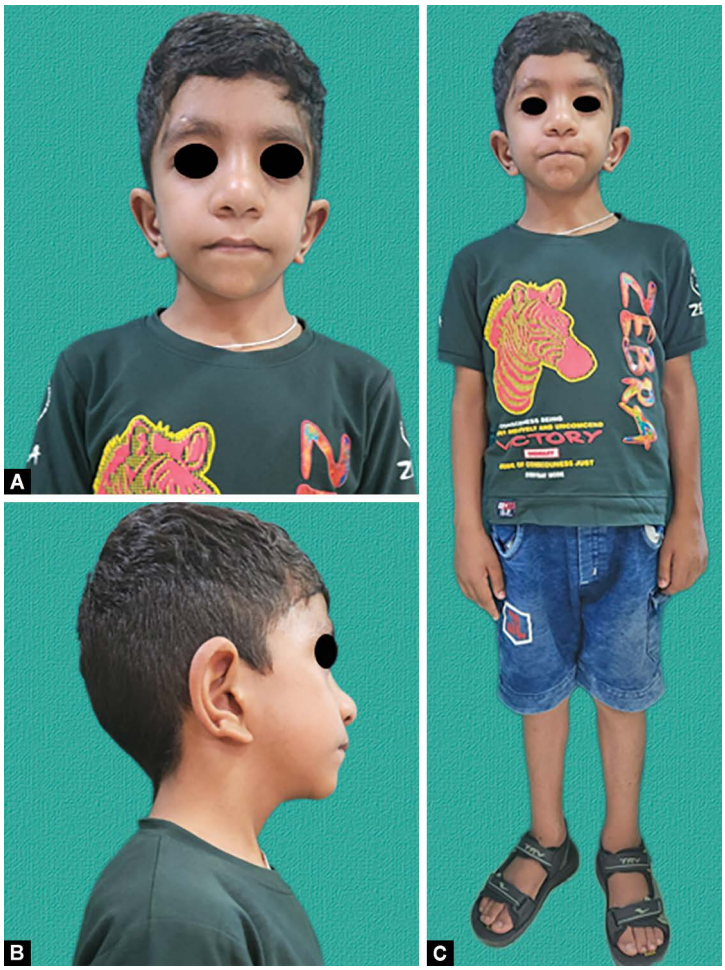
stunted growth, so he was referred to an endocrinologist, after which an investigation was done, and it revealed bone age of 4.5–5 years at a chronological age of 5.5 years. The patient was initiated on growth hormone therapy and is actively on injection somatotropin (Genotropin or Headon, 1.8 IU) since the last 6 months for short stature. Electrocardiogram (ECG) was done and no abnormal findings were recorded. Family history revealed that the mother developed bluish spots on and off. It was also reported that it was a nonconsanguineous marriage.

Extraoral examination showed associated features of NS like down-slanting palpebral fissures, hypertelorism, a flat nasal bridge, low-placed ears, a long philtrum, and downturned lips. The patient also presented with short stature (Fig. 1).

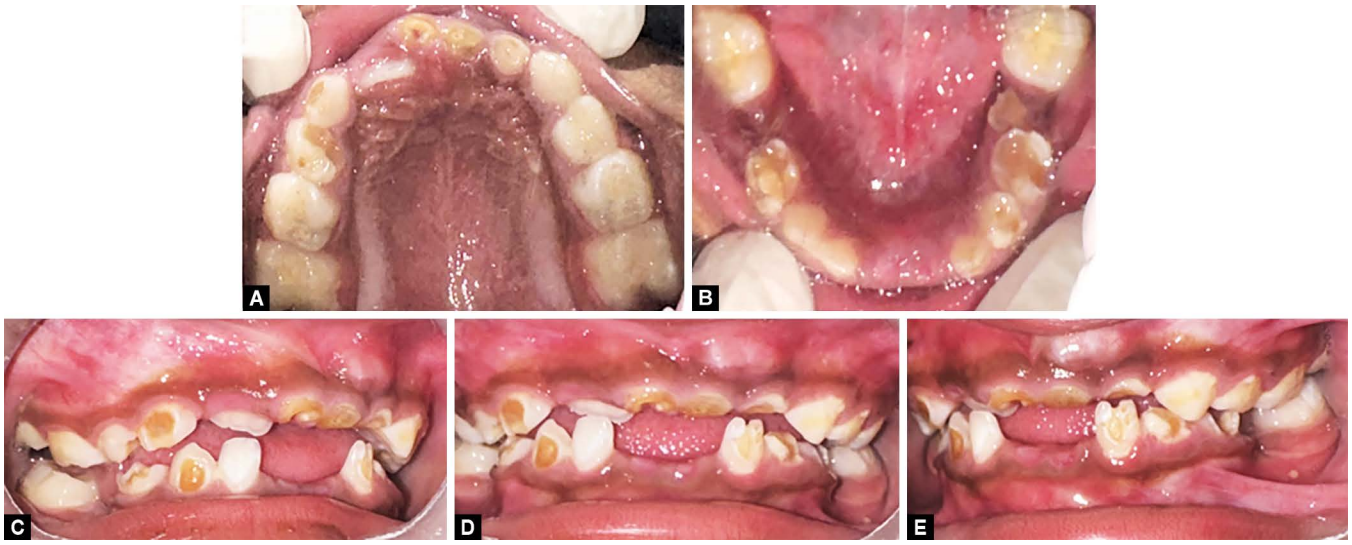
It was the patient's first dental visit. The patient had a history of poor oral hygiene and prolonged bottle feeding, leading to multiple carious lesions.

Intraoral examination revealed a mixed dentition stage. Present teeth included: 16, 55, 54, 53, 11, 51, 61, 62, 63, 64, 65, 26, 46, 85, 84, 83, 82, 72, 73, 74, 75, and 36. Root stumps were noted for teeth 51, 61, 62, 75, and 85, which were nonrestorable. Smooth-surface caries was noted with respect to 53, 63, 72, 73, and 83. Additionally, dental caries was present with respect to 54, 64, 74, and 84. Deep pit and fissure morphology was noted on all the permanent first molars (Fig. 2).

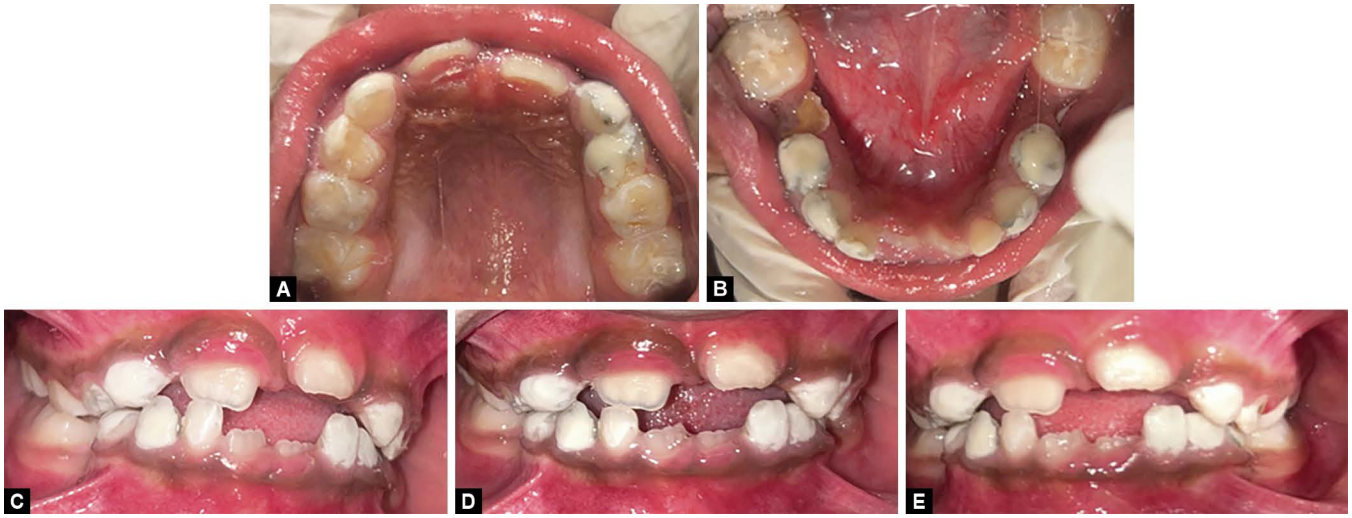
Extraction was planned with respect to teeth 51, 61, and 62 after obtaining consent from the pediatrician, as this addressed the patient's chief complaint. Prior to the dental procedure, the patient underwent a thorough cardiovascular evaluation, and written clearance was obtained from the pediatric cardiologist confirming hemodynamic stability for dental extractions under local anesthesia. He was advised for blood investigation: CBC, PT, aPTT, and international normalized ratio (INR). The CBC was normal. PT was 11.8 seconds (within normal range: 9.7–13.7 seconds), and INR was 0.94 (within normal range: 0.8–1.2). However, aPTT was slightly prolonged at 34.50 seconds (normal range: 23.5–32.5 seconds). So, the child was advised to proceed for extraction with Tranexa 500 mg, half a tablet, half an hour before extraction. Extraction was done under local anesthesia (2% Lidocaine with 1:1,00,000 epinephrine) with respect to 51, 61, and 62 using an infiltration technique, avoiding regional blocks to minimize the risk of hematoma formation. Aspiration was carefully performed prior to injection. Atraumatic extraction technique was employed using minimal force and appropriate instruments to preserve surrounding tissues. Teeth 51, 61, and 62 were extracted sequentially. After extraction, gelatin sponge (Gelfoam) impregnated with tranexamic acid solution was placed in each socket. Firm pressure was applied to the extraction sites with sterile gauze for 10–15 minutes. The patient was monitored closely in the department for 1-hour postprocedure for any signs of active bleeding. Postextraction paracetamol was prescribed for pain relief. NSAIDs (e.g., ibuprofen) were strictly avoided due to their antiplatelet effects. Tablet Augmentin



Figs 1A to C: Extraoral photographs: (A) Frontal view of the child showing hypertelorism, a flat nasal bridge, a long philtrum, and downturned lips; (B) Lateral view showing low-placed ears; (C) Short stature of the patient



Figs 2A to E: Intraoral view showing early childhood caries: (A) Maxillary occlusal view showing root stumps with respect to 51, 61, and 62. Smooth-surface caries with respect to 53 and 63. Dental caries with respect to 54; (B) Mandibular occlusal view showing root stumps with respect to 75. Smooth-surface caries with respect to 72, 73, and 83. Dental caries with respect to 74 and 84; (C) Right lateral view; (D) Central view; (E) Left lateral view



Figs 3A to E: Postoperative photographs: (A) Maxillary occlusal intraoral view showing uneventful healing of extraction site, silver modified atraumatic restorative treatment with respect to 53, 54, 63, and 64. Pit and fissure sealant done with respect to 16 and 26; (B) Mandibular occlusal view: silver modified atraumatic restorative treatment with respect to 72, 73, 74, 83, and 84. Pit and fissure sealant done with respect to 36 and 46; (C) Right lateral view; (D) Central view; (E) Left lateral view

375 mg was also prescribed thrice a day for 5 days.[6] Postoperative instructions were given. Gentle rinsing with chlorhexidine mouthwash was advised starting 24 hours postextraction. Brushing near the extraction sites was to be avoided for the first few days. A soft, cold diet was recommended for the initial 24–48 hours. Detailed instructions were given to the parents regarding signs of bleeding, pain management, diet, and oral hygiene. The patient was advised to take Tranexa 500 mg, half a tablet, if any significant bleeding occurred. A follow-up appointment was scheduled after 1-week postextraction to assess wound healing. Extraction sites with respect to 51, 61, and 62 showed uneventful healing (Fig. 3). As the patient showed high caries index, all the permanent molars were sealed with fluoride-releasing resin-based pit and fissure sealant. Silver diamine fluoride application was done with respect to 53, 54, 63, 64, 72, 73, 74, 83, and 84, followed by glass ionomer restoration. Since the permanent upper right central incisor was erupting in crossbite, tongue blade therapy was recommended (Fig. 4).

Discussion

This report underscores the dental management approach for a patient with known VWD and NS undergoing dental extraction. The main concern while treating a patient with VWD is risk of excessive bleeding during invasive procedure. The use of antifibrinolytic agents such as tranexamic acid is advocated for preventing clot lysis and achieving hemostasis once extraction[7] procedure is done. 43% of patients often have factor XI deficiency, platelet dysfunction, or any other coagulopathies, which result in higher incidence of bleeding disorder.[8]

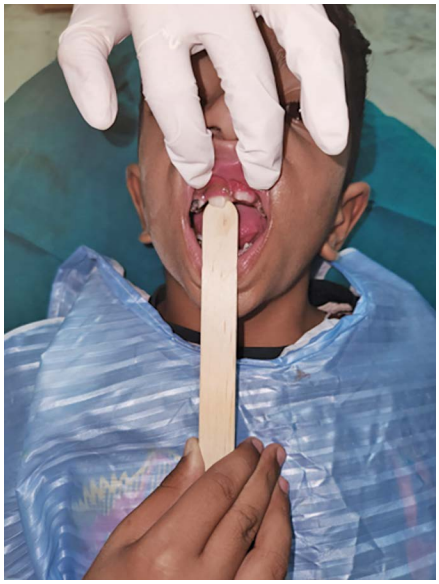


Fig. 4: Demonstration of tongue blade therapy

While the primary bleeding risk in this patient was attributed to VWD, the potential for additional coagulation defects associated with NS necessitated a thorough preoperative hematological assessment.[8] The cardiac history given by parents was nonsignificant, but patient was asked for cardiological clearance to prevent any cardiac complications during the procedure to ensure patient's safety. These types of cases demand multidisciplinary approach between pediatric dentistry, pediatric hematology, and pediatric cardiology for its successful management.[9] Atraumatic extraction and meticulous use of local hemostasis is of paramount importance. Local hemostatic agents (gelatin sponge with tranexamic acid) and pressure pack combination resulted in more beneficial mechanical and pharmacological aid for clot formation and stability.[10,11]

Limitations

The study is limited by its single-case design, lack of control group, and limited long-term follow-up, which may affect generalizability.

Conclusion

Von Willebrand disease and NS patients require a well-planned, individualized, and tailored treatment approach. For optimal clinical outcomes in such cases, a thorough preoperative history, use of hemostatic agents, atraumatic extraction technique, and close postoperative monitoring are required. This case highlights the importance of customized dental management plan for patients with hematological and systemic diseases, to ensure minimum risk of bleeding complications.

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