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Retrobulbar haemorrhage in a Factor XIII deficiency patient a case report

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Abstract : Factor XIII deficiency is a rare bleeding disorder with an incidence of one in 2.5 million population. It presents with prolonged umbilical cord stump bleeding during neonatal period, delayed soft tissue bruising, mucosal bleeding and with life- threatening intra cranial haemorrhage. Here report a 14 years old boy, known case of factor XIII deficiency presented with acute proptosis and defective vision in the Right eye following Road Traffic Accident, diagnosed to have Retrobulbar haemorrhage, who was immediately underwent for hematoma evacuation with lateral canthotomy under general anesthesia and he received 6 units of cryoprecipitate before and after the procedure as per Hematologist instruction.Postoperatively, the retrobulbar hemorrhage regressed and vision improved to 6 6p

Keyword : Factor XIII deficiency, Retrobulbar Hemorrhage, Proptosis

Case Report

A 14 years old boy, known case of factor XIII deficiency, presented with protrusion of Right eye for past 3 days, following fall from two wheeler. He had defective vision with associated photophobia, watering and pain in the right eye. He was diagnosed as a case of factor XIII deficiency in his first year of age when he developed haematoma at the vaccination site. He had past history of dental bleeding and frequent soft tissue haematoma following trivial injuries, for that he had multiple episodes of cryoprecipitates transfusion. His elder sibling died in his first week of age due to intra cranial haemorrhage. On examination he was conscious, oriented to time, place and person with stable vitals. Systemic examination was normal. On ocular examination, his Right eye vision was counting fingers at 1 meter. There was an eccentric proptosis of downward displacement with periorbital oedema and ecchymosis. The conjunctiva had grade4 chemosis with dense sub conjunctival haemorrhage. The Cornea showed oedema with inferior exposure keratopathy (Figure:1).

Figure:1 showing Acute proptosis with dense SCH

Pupillary reaction showed relative afferent pupillary defect with clear lens. All extra ocular movements were restricted. On fundus examination, view was hazy due to corneal oedema and exposure keratopathy. Disc and vessels hazily seen. On examination of left eye, visual acuity was 6/6, anterior segment and posterior segment were normal. The patient was clinically diagnosed to have Retrobulbar haemorrhage in Right eye and subjected to urgent CT ORBIT and he was started on systemic steroids (T.Prednisolone 1mg/kg), T.Diamox 250mg twice daily with Topical antibiotics, lubricants with lid taping. CT showed intraconal hematoma and extraconal haematoma in the superior aspect of orbit. (Figure:2&3a,b))

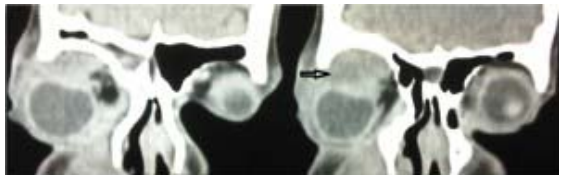

Fig:2 CT orbit Coronal view shows Haematoma in the superomedial aspect of right orbit

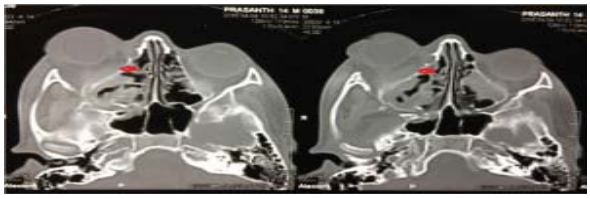

Fig:3a Axial view showing Intraconal hematoma in the right orbit

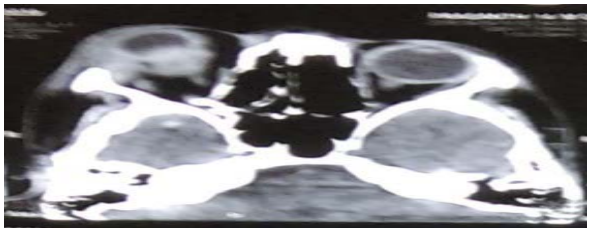

Figure:3b Axial view showing intraconal hematoma in the right orbit

After consulting with Haematologist, the patient was posted for immediate haematoma evacuation with cover of cryoprecipitates .Under general anaesthesia , 6ml of blood was aspirated from the superomedial aspect of right orbit with lateral canthotomy.(Figure:4).6 units of cryoprecipitates were given pre and post procedure as per haematologist opinion.(7,3)

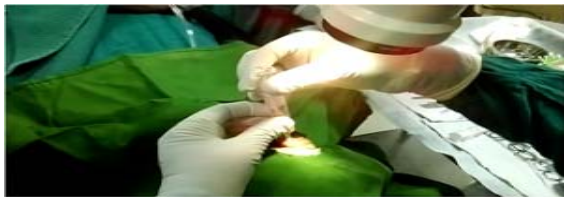

Figure:4 shows intra operative picture showing evacuation of blood

In the immediate post operative period, patient was carefully monitored for any increase in size of proptosis, pupillary reaction and extra ocular movements and he was advised to continue systemic steroids, topical antibiotics and lubricants. On the 5th post operative day, his vision improved to 4/60, the size of proptosis and chemosis reduced. Minimal extra ocular movements were appreciated. Inferior exposure keratopathy was healing. On 3rd week of post operative period, his vision improved to 6/9. There was no proptosis and chemosis regressed. Extra ocular movements were full. Exposure keratopathy healed. (Figure: 5)

Figure: shows Regressed proptosis with healing exposure keratopathy On 6th week of post operative period his vision was 6/6p.(Figure:6)

Figure:6 Shows regressed proptosis with healed proptosis

Discussion:

Congenital factor XIII deficiency is a rare autosomal recessive disorder usually associated with severe bleeding diasthesis with the incidence rate of one in 2.5 million population.(3)

Pathophysiology:

Factor XIII is a plasma transglutaminase that catalyzes the final step in the coagulation cascade, cross-linking the loose fibrin polymer into a highly organized structure.(2) The zymogen FXIII (pFXIII) circulates in plasma as a tetramer (A2B2), consisting of two catalytic A subunits (FXIII-A) and two carrier/protective B subunits (FXIII-B). The zymogen FXIII is activated to FXIIIa by thrombin and ca2+ (2,4) Factor XIIIa catalyzes the formation of covalent bonds between glutamine and lysine residues on the fibrin a and g chains, enhancing the mechanical strength of the fibrin polymer.(4) Congenital FXIII deficiency can be due to defects in either FXIII-A genes (also known as type 2 defect) or FXIII-B genes (type 1 defect). FXIII B subunit gene mutation occurs infrequently of about <5% . FXIII A gene mutation is the usual occurrence with severe bleeding manifestations.(4,6,7)

Clinical presentations:

Bleeding from the umbilical cord stump within the first day to weeks of life is a characteristic sign that occurs in 80% of the individuals. Delayed bleeding (i.e., 12-36 h) after trauma or surgery is pathognomonic of factor XIII deficiency.(1,8) CNS haemorrhage, soft tissue bleeding , hemoarthoses which occur spontaneously or after minor trauma with delayed wound healing are the other manifestations. Recurrent spontaneous abortions are common in women with factor XIII deficiency.(2,4,9)

Diagnosis:

The standard laboratory clotting tests, such as prothrombin time, activated partial thromboplastin time, fibrinogen level, platelet count and bleeding time and international normalized ratio (INR) are normal in factor XIII (FXIII) deficiency.(3) Assessment of clot stability is the most common screening test for factor XIII deficiency. In the presence of factor XIII, the clot is stable for more than 24 hours; in its absence, the clot dissolves in minutes to hours. (4,8,9) The clot solubility test is only sensitive at very low levels of FXIII (zero or very close to zero) and will be normal if the FXIII activity level rises upto 1–3% . If the clot solubility test result is positive for lysis, quantitative analysis of FXIII activity should be done. Genetic analysis and specific FXIII mutational analysis is performed to aid in family counselling, prenatal screening.(9)

Treatment:

Plasma, cryoprecipitate, and factor XIII (FXIII) concentrates have been used for replacement of factor XIII and the treatment of bleeding.(4) Recombinant factor XIII-A2 (Tretten) was approved by the FDA in December 2013 and monthly 35 IU/kg r FXIII A-subunit injections significantly decreased the number of treatment-requiring bleeding episodes.(6) Around 3-5% of Factor XIII are usually sufficient to prevent spontaneous bleeding and Scheduled factor XIII (FXIII) replacement every 4-6 weeks maintains factor XIII levels above the critical threshold for spontaneous bleeding and allows patients to participate in regular activities. Prophylactic therapy with factor XIII concentrate 10-20 U/kg every 4-6 weeks or recombinant FXIII A-subunit (35 IU/kg/month IV) provides adequate plasma levels in most patients. For surgical procedures, patients should receive factor XIII concentrate immediately before surgery to ensure optimal hemostasis and wound healing.(8,5)

Prognosis:

Eventhough there is a lifelong risk of bleeding with FXIII deficiency, the prognosis is excellent because of the good response to treatment with FFP, cryoprecipitate or plasma-derived FXIII concentrate.(9)

Conclusion:

This case is an inherited FXIII deficiency diagnosed at the age of one year with various bleeding manifestation. This case is reported for its rare presentation and to emphasise the importance of timely intervention to save the vision.

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