Frequency & clinical spectrum of rare bleeding disorder in Pakistan, A multi centre study.

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Background

According to the recent "Pakistan Demographic and Health Survey (DHS) ", the two-third of marriages are consanguineous[1]. Rare bleeding disorders (RBD) prevalence is high in those countries where consanguinity is normally practiced and pose significant clinical & social problem[2]. It is generally transmitted as an autosomal recessive trait and divided into hereditary or acquired. It can be defects of vasculature, platelet (Deficiencies of membrane glycoprotein, disorders of storage granules and familial thrombocytopenia) and coagulation proteins (Factor II, V, VII, X, XI, XII, XIII, Combined V and VIII, Multiple clotting factor deficiencies)[3]. Lack of knowledge about natural history, diagnosis and optimal management due to less prevalence in the population. Most of the labs label these patients as hemophilia or misdiagnose. RBD's are further divided into type I and II deficiencies[4]. Frequency of these disorders varies from population to population exact figures are unknown especially in our region. Bleeding pattern may considerably vary between affected individuals[5].

Objective

To estimate the incidence of rare bleeding disorder and their clinical presentation at multiple hematology centers in Pakistan.

Study Design And Method

Descriptive and cross sectional

Setting: Total 732 patients were evaluated for bleeding tendency at the multiple centers & 172(23.4%) were diagnosed to have rare bleeding disorders.

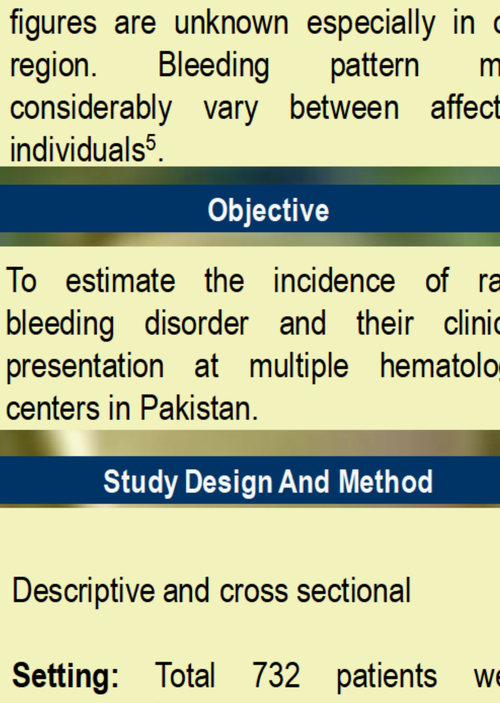


Figure 1: Multi centre studies

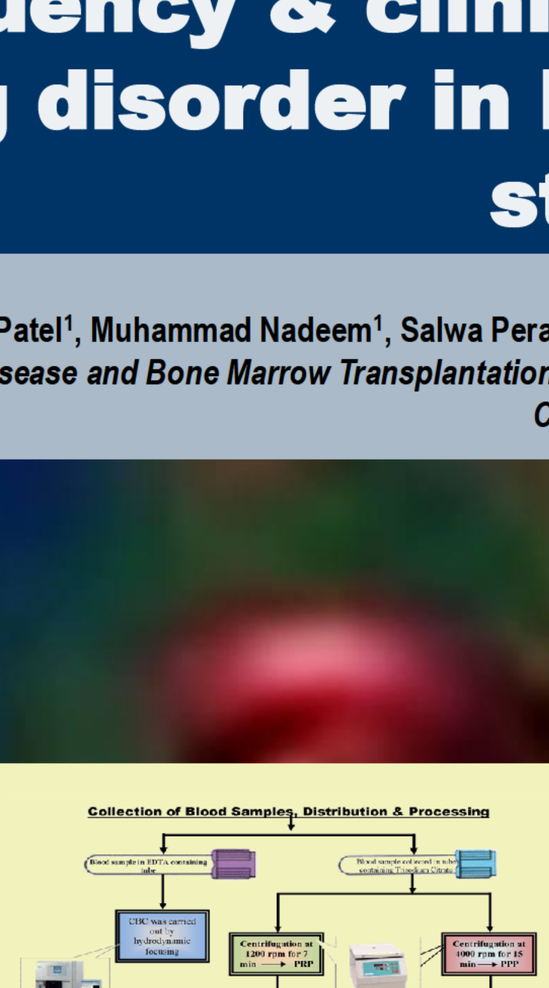


Figure 2: Collection , distribution and analysis of blood specimen

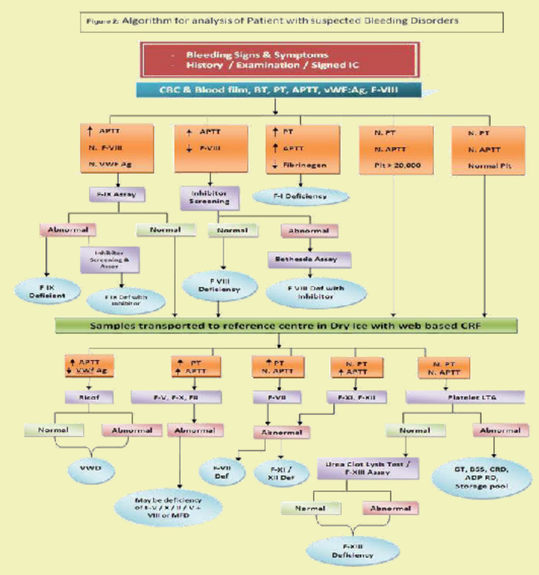


Figure 3: Algorithm for analysis of Patient with suspected Bleeding Disorders

Results

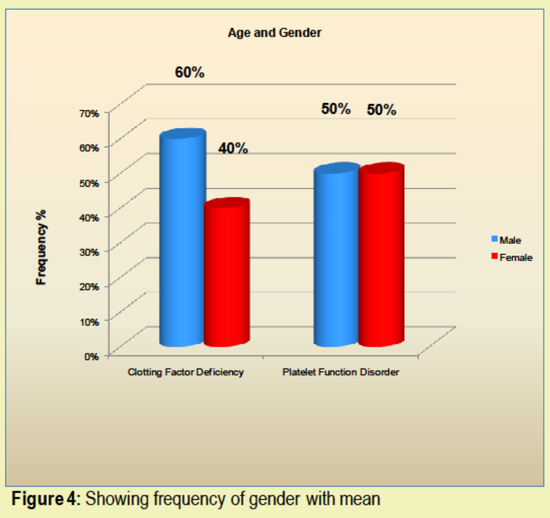


Figure 4: Showing frequency of gender with mean age

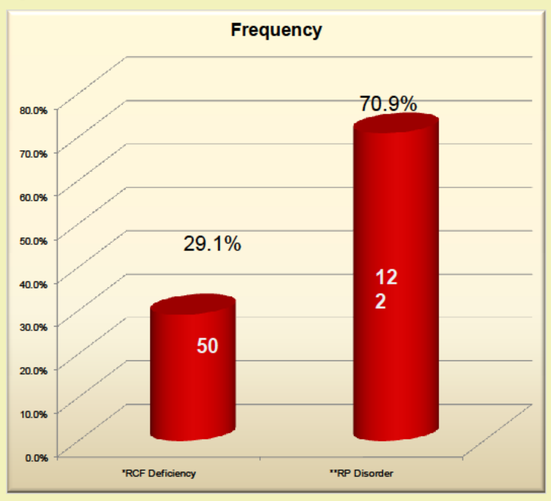


Figure 5: Showing frequency of Rare bleeding Disorders
*RCF Deficiency = Rare Clotting Factor Deficiency
**RP Disorder = Rare Platelet Disorder

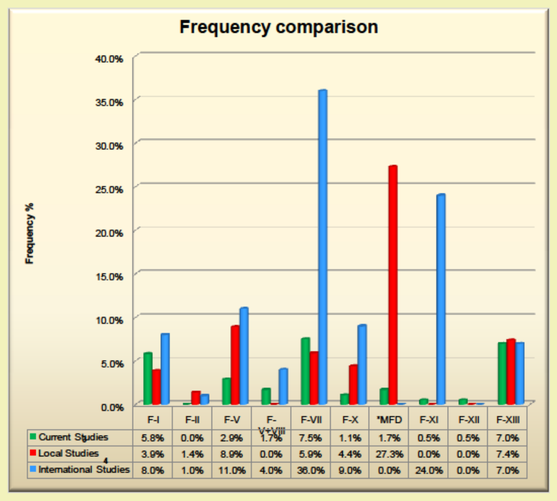


Figure 6: Showing frequency comparison of current, local & international studies

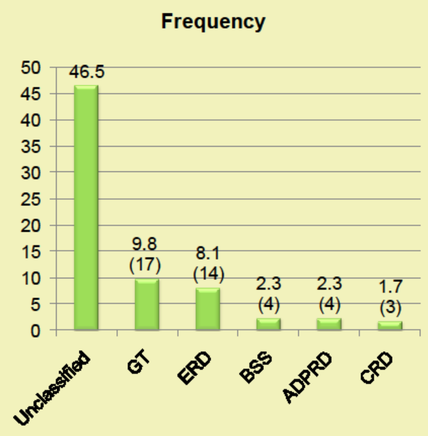
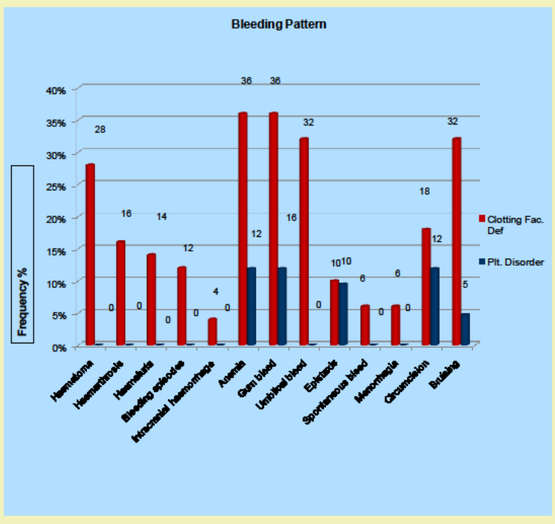


Figure 7: Showing frequency of rare platelet disorder
[1]GT = Glanzmann Thrombasthenia [2]ERD = Epinephrine Receptor Defect [3]BSS = Bernard Soulier Syndrome
[4]ADPRD = ADP Receptor Defect [5]CRD = Collagen Receptor Defect [6]Tot. Plt. Dis = Total Platelet Disorder



Conclusion

Current study is showing the frequency of rare bleeding disorder of about 172 (23.4%). Out of these, 50 (29.1%) were diagnosed as rare clotting factor deficiency and 122 (70.9%) as rare platelet function disorder.

By comparing our results with other local and international studies we observed that the frequency of these autosomal recessive rare bleeding disorders is variable. These disorders are more frequent in the population where consanguineous marriages are frequent including our population[5].

Our results are not exactly true reflection of frequency of RBD especially in case of platelet disorders because study was not totally randomized or hospital based.
The total number of patients with rare bleeding disorders studied by us is limited and large number of patients remains undiagnosed due to limited resources. There is a need of more studies in all parts of our country to estimate the actual frequency of these disorders.

Future Directions

- Large scale studies to estimate accurate frequency.
- Analytical expertise for the assessment of more platelet function disorders.
- Analytical expertise for specific gene mutations.

Acknowledgment

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Disclosure of Interest : Non declared

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