J Blood Disord Transfus 2017, 8:2(Suppl)
http://dx.doi.org/10.4172/2155-9864-C1-023



## Hematological parameters of three freshwater stingray species (chondrichthyes: Potamotrygonidae) in the middle Rio Negro, Amazonas state

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This paper aimed to study and compare the hematology of newborns, young, subadults, adult males, adult females and pregnant females of *Potamotrygon wallacei* (cururu stingray), *Potamotrygon motoro and Paratrygon aiereba*. Newborn cururu stingrays had lower red blood parameters than those of other development stages. Thrombograms and leukograms showed a conservative pattern between development stage, sexual dimorphism and pregnancy. *In P. motoro* and *P. aiereba*, variables relating to red blood parameters, biochemistry and leukograms showed little variation between the species' biological characteristics, thus showing that these variables are not good criteria for differentiating them within the same species. In conclusion, the development stage is an important factor for differentiating hematological properties in the cururu stingray, while this has not been observed in *P. motoro* and *P. aiereba* stingrays.

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## Novel molecular changes in Saudi patients with familial hemophagocytic lymphohistiocytosis

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**Background**: Familial hemophagocytic lymphohistiocytosis (FHL) in different ethnicities has been described in the literature, but this is the first report from Saudi Arabia describing the novel mutations present in *FHL* genes.

**Methods**: 87 patients diagnosed with *FHL* from January 1995 to December 2014 at King Faisal Specialist Hospital and Research Centre was screened for HLH-associated genes. Their clinical and biochemical profiles were retrospectively captured. DNA from peripheral blood were used for mutation detection in various *HLH* genes- *PRF1, UNC13D, STX11, STXBP2, LYST, rab27A, SH2D1A* and *XIAP* by PCR-sequencing method. We report herein those with novel molecular changes.

**Results**: Biallelic mutations were identified in 66 patients (75.86%) in whom 18 (27.3%) patients were found to harbor 10 novel mutations distributed among five HLH-associated genes. *STXBP2* mutations were identified in the majority of patients (38%). All mutations were found to be damaging and disease. 10 patients with *UNC13D* had four novel mutations, two of which resulted in a stop codon. The most prevalent mutation is c.3048_3049insC (p.E1017RfsX8) was found in six patients. One patient had a novel missense mutation (c.862 T>C, p.W288R) in *STXBP2* gene. Another *STX11* mutation (601_602ins C, p. Q140Pfs*46) was found in one patient. Four novel mutations were found in seven patients in other genes *(LYST and rab27A)*. The novel molecular changes and their associated clinical characteristics were shown. Parent consanguinity and history of siblings with *HLH* were observed in 77% and 26% of patients, respectively. Furthermore, a tribal and geographical pattern was clearly found in patients harboring *STX11, STXBP2* and Unc13D mutations. *STXBP2* mutations are the most prevalent among Saudi FHL patients.

**Conclusion**: In more than a quarter of mutations in Saudi patients with FHL are novel. Furthermore, in quarter of our patients, no molecular defects were identified. This indicates that there are still more mutations to be discovered and also the possibilities of deep intronic mutations and other genetic aberrations cannot be definitely excluded. A high rate of consanguineous marriages and endogamy is seen in Saudi Arabians', and is present in large groups. A tribal and geographical pattern was clearly observed. Though the treatment is standardized for HLH, the impact of ethnicity and race on the severity and outcome may warrant further investigation.

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