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Effect of GAA expansion on Iron Copper metabolism and cell free nucleic acids (cfNA) levels in plasma of Friedreich's ataxia (FRDA) patients and its co relation with the FARS

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Friedreich's ataxia is characterized by the high iron and copper deposits in the brain and cardiac cells which triggers the cellular and axonal death causing increased plasma levels of the circulating nucleic acids in the patients. A quantitative study was done and the plasma levels of cell free Nucleic acids, and trace elements iron and copper were assessed and correlated with the GAA repeat numbers and frataxin levels in patients. 25 FRDA patients and 25 controls from Northern India were included. Iron and Copper level assessment were done by Nitro PAPS and Dibrom PAESA method, respectively. Fluorescent dye-based Qubit 3.0 Fluorometer was used for cfNAs quantifications. Iron (Fe²⁺) and Copper (Cu²⁺) levels were found to be significantly

decreased in patients (Cu; mean \pm SD(range) 8 \pm 5(2-16); Fe mean \pm SD(range) 5 \pm 3(15-13) compared to controls(Cu; mean \pm SD(range) 13 \pm 8(12-29); Fe mean \pm SD(range) 16 \pm 7(4-26) whereas free cfDNA levels were found to be higher in patients (mean \pm SD(range) 73 \pm 35(2-16)) compared to controls (mean \pm SD(range) 34 \pm 0.3(9-6)). There was a significant positive correlation between GAA repeat numbers and cfDNA & frataxin level and Fe and Cu levels. Significant inverse correlation was established between GAA repeat numbers and Fe and Cu levels & frataxin and cfNA.

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