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Houyem Ouragini [1], Emna Bouatrous [1], Sonia Nouira [1], Hamza Dallali [2], Samia Rekaya [1,3], Dorra Chaouachi [1], Monia Ouederni [1,3], Menif Samia [1]

[1] Laboratory of Molecular and Cellular Hematology,LR16IPT07, Institut Pasteur de Tunis, University of Tunis ElManar, Tunis, Tunisia;
[2] Laboratory of Biomedical Genomics and Oncogenetics, Institut Pasteur de Tunis, University of Tunis ElManar, Tunis, Tunisia;
[3] Pediatric Immuno-Hematology Unit, Bone Marrow Transplantation Center, Tunis, Tunsia.

Neurological impairment in hemoglobin disorders: first cases in Tunisia

Hemoglobin disorders are among the most common inherited diseases worldwide. Their clinical manifestations range from anemia to more severe forms associated with neurological impairments. These complications can result as secondary consequences of the disease's clinical manifestations, or be directly linked to genetic mutations. In this study, we present two families with neurological impairments who were referred to us for complementary hematological and biochemical analyses. Complete blood count, methemoglobin level, and methemoglobin reductase activity were assessed. Molecular analyses were performed using whole-exome sequencing, and segregation of the identified mutations was confirmed with direct sequencing. Their pathogenicity and conservation were evaluated using various bioinformatics tools. Clinical and hematological findings suggested X-linked alpha thalassemia/mental retardation syndrome in the first family and recessive congenital methemoglobinemia type II in the second. This was confirmed by the identification of pathogenic mutations ATRX: p.Arg2131Gln and CYB5R3: p.Ala179Thr, respectively. Although these variants have been previously reported worldwide, they were identified for the first time in our population. Our results contribute to the understanding of the pathogenesis of these rare disorders and provide a basis for diagnosis, treatment, and genetic counseling. The mechanisms by which these mutations contribute to neurological symptoms are discussed.

Keywords
ATR-X Syndrome; Recessive Congenital Methemoglobinemia Type II; neurological disease; mutation; WES; diagnosis

Biography
University, where her master's and PhD work focused on molecular investigation of epidermolysis bullosa, a rare genodermatosis. She is currently Associate Professor at the Laboratory of Molecular and Cellular Hematology, Pasteur Institute of Tunis. Her research interests include epigenetic regulation of HbF expression, a key modulator of disease severity in hemoglobinopathies (SCD, and β-thalassemia), and the phenotypic/molecular characterization of rare erythroenzymopathies. She has published in peer-reviewed journals, presented at international conferences, and is actively involved in teaching and supervising postgraduated students.

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