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Clinical Classification, Diagnosis and Treatment for Thalassemia

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Message from the Guest Editor

Rare anaemias (RAs), ORPHA108997, include up to 132 rare and ultra-rare haematological conditions, representing a highly heterogeneous group of disorders. These are characterized by anaemia of variable degree, from mild forms to life-threatening chronic blood-transfusion dependence, and by complex and often unexplained genotype-phenotype correlations. More than 80% of RA are genetic disorders caused by mutations in more than 70 genes controlling red blood cell (RBC) production and structure. These mutations lead to alterations in haemoglobin (Hb) structure or synthesis, RBC maturation and differentiation, cell membrane structure, and enzyme deficiencies. The balance between haemolysis (mainly in the spleen) and erythropoiesis explains the severity of the anaemia and the patient's ability to respond to treatment(s). In this context, differential diagnosis, prognosis and patient stratification are often difficult. Some studies have already demonstrated the usefulness of the targeted-NGS (t-NGS) approach in the investigation of specific subtypes of RA.

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