Role of different phenotypic groups of thalassemia major patients studied by CMR

Laura Pistoia¹, Antonella Meloni¹, Paolo Ricchi², Rosamaria Rosso², Aurelio Maggioª, Domenico D’Ascola², Zelia Borsellino³, Saveria Campisi¹, Maria Grazia Roberti⁴, Lorella Pitrolo⁶, Stefania Renne⁷, Alessia Pepe¹


Introduction. Beta thalassemia major (β-TM) displays a great deal of phenotypic heterogeneity, not fully investigated in terms of cause-effect.

Aim. We aimed to detect if different phenotypic groups could be related to different levels of cardiac impairments, evaluated by cardiovascular magnetic resonance (CMR).

Methods. We studied retrospectively 671 β-TM patients (age 30.1 years, 52.9% females) enrolled in the Myocardial Iron Overload in Thalassemia (MIOT) network. Myocardial iron overload was assessed by using a multislice multiecho T2* approach. Cine sequences were obtained to quantify biventricular functional parameters.

Results. Three groups of patients were identified: heterozygotes (N=279), homozygotes β⁺ (N=154), homozygotes β⁻ (N=238). No significant differences for sex, age and haemato-chemical parameters were found among the groups. Transfusional needs resulted significantly lower in the homozygous β⁺ patients than the heterozygous (34.7±11.3 U vs 38.0±12.7 U, P<0.05) and the homozygous β⁻ patients (34.7±11.3 U vs 41.6±12.7, P<0.0001). After adjusting for the transfusional requirements, the homozygous β⁺ group showed significantly higher global heart T2* values than the homozygous β⁻ group (32.4±10.4 ms vs 26.2±13.0 ms, P<0.01) and a significantly lower number of segments with T2*<20 ms than both the heterozygous (3.0±5.0 vs 4.7±6.1, P<0.05) and the homozygous β⁻ group (3.0±5.0 vs 5.9±6.6, P<0.01) groups; the number of patients with a global heart T2* value<20 ms was significantly lower in the homozygous β⁺ group when compared to the other groups (homozygous β⁺ vs heterozygous = 14.3% vs 26.9%, P<0.05; homozygous β⁺ vs homozygous β⁻ = 14.3% vs 34.0%, P<0.01). Moreover, after adjusting for cardiac iron, the homozygous β⁺ group showed a lower number of patients with a pathological left ventricular ejection fraction (LVEF) than the heterozygous and homozygous β⁻ groups (14.3% vs 24.2%, P<0.05 and 14.3% vs 27.2%, P<0.05).

Conclusions. The homozygous β⁺ TM patients showed less myocardial iron overload and a concordant lower frequency of systolic heart dysfunction and cardiac remodelling. These data support the knowledge of the different phenotypic groups in the clinical and instrumental management of β-TM patients.