

**ID: 109**

**Area Clinica**

**Poster**

*Parole chiave:* Thalassemia major, cardiac iron overload, cardiovascular magnetic resonance, phenotypic group

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

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**Introduction.** Beta thalassemia major ( $\beta$ -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  $\beta$ -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  $\beta$ + (N=154), homozygotes  $\beta$ <sup>o</sup> (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  $\beta$ + patients than the heterozygous ( $34.7 \pm 11.3$  U vs  $38.0 \pm 12.7$  U,  $P < 0.05$ ) and the homozygous  $\beta$ <sup>o</sup> patients ( $34.7 \pm 11.3$  U vs  $41.6 \pm 12.7$ ,  $P < 0.0001$ ). After adjusting for the transfusional requirements, the homozygous  $\beta$ + group showed significantly higher global heart T2\* values than the homozygous  $\beta$ <sup>o</sup> group ( $32.4 \pm 10.4$  ms vs  $26.2 \pm 13.0$  ms,  $P < 0.01$ ) and a significantly lower number of segments with T2\* < 20 ms than both the heterozygous ( $3.0 \pm 5.0$  vs  $4.7 \pm 6.1$ ,  $P < 0.05$ ) and the homozygous  $\beta$ <sup>o</sup> ( $3.0 \pm 5.0$  vs  $5.9 \pm 6.6$ ,  $P < 0.01$ ) groups; the number of patients with a global heart T2\* value < 20 ms was significantly lower in the homozygous  $\beta$ + group when compared to the other groups (homozygous  $\beta$ + vs heterozygous = 14.3% vs 26.9%,  $P < 0.05$ ; homozygous  $\beta$ + vs homozygous  $\beta$ <sup>o</sup> = 14.3% vs 34.0%,  $P < 0.01$ ). Moreover, after adjusting for cardiac iron, the homozygous  $\beta$ + group showed a lower number of patients with a pathological left ventricular ejection fraction (LVEF) than the heterozygous and homozygous  $\beta$ <sup>o</sup> groups (14.3% vs 24.2%,  $P < 0.05$  and 14.3% vs 27.2%,  $P < 0.05$ ).

**Conclusions.** The homozygous  $\beta$ + 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  $\beta$ -TM patients.