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Genotypic groups as risk factor for cardiac MR abnormalities and complications in thalassemia major

Authors:
A Pepe1, A Meloni1, S Salvadori2, A Vallone3, S Renne4, G Peritore5, P Preziosi6, A Riva7, M Missere8, PP Bitti9, V Spadola10, V Positano1, L Pistoia1, 1Fondazione Toscana Gabriele Monasterio - Pisa - Italy, 2Institute of Clinical Physiology, CNR - Pisa - Italy, 3Garibaldi Hospital - Catania - Italy, 4P.O.Giovanni Paolo II - Lamezia Terme - Italy, 5Ospedale Civico - Palermo - Italy, 6Sandro Pertini Hospital - Rome - Italy, 7OSP. SS. Annunziata ASL Taranto - Taranto - Italy, 8Centro di Ricerca e Formazione ad Alta Tecnologia nelle Scienze Biomediche - "Giovanni Paolo II" - Campobasso - Italy, 9San Francesco Hospital - Nuoro - Italy, 10Azienda Ospedaliera Civile - O.M.P.A. Ragusa - Ragusa - Italy,

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Introduction. Beta thalassemia major (β-TM) displays a great deal of genotypic heterogeneity, not fully investigated in terms of cause-effect. This prospective and multicentre study aimed to detect if different genotypic groups could predict the development of cardiovascular magnetic resonance (CMR) abnormalities and cardiac complications (CC).

Methods. We considered 708 β-TM patients (373 females, 30.05±9.47 years), consecutively enrolled in Myocardial Iron Overload in Thalassemia (MIOT) network. Data were collected from birth to the first CMR imaging scan. Myocardial iron overload was assessed by the multislice multiecho T2* technique. Biventricular function parameters were quantified by cine images. Late gadolinium enhancement (LGE) images were acquired to detect myocardial fibrosis.

Results. On the basis of the type of gene mutation, three groups of patients were identified: homozygotes β+ (N=158), compound heterozygotes β+/β° (N=298) and homozygotes β° (N=252). Table 1 shows the effect of genotype group on the development of different cardiac outcomes. Compared to the milder genotype group homozygotes β+, the other two groups showed a significantly higher risk of myocardial iron overload (MIO) and left ventricular dysfunction. We recorded 90 (13.0 %) cardiac events: 46 heart failures (HF), 38 arrhythmias (33 supraventricular, 3 ventricular and 2 hypoinetic) and 6 pulmonary hypertensions (PH). No prospective association was detected between genotype group and HF and PH. The homozygous β° group showed a significantly higher risk of arrhythmias than the homozygous β+ group and at the limit of significance than the compound heterozygotes. Globally, homozygotes β° showed a significantly higher risk of CC than homozygotes β+.

Conclusion. Different genotypic groups predict the development of MIO, left ventricular dysfunction, arrhythmias and CC in β-TM patients. These data support the knowledge of the different genotypic groups in the clinical management of β-TM patients.
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1 Fondazione Toscana Gabriele Monasterio - Pisa - Italy, 2 Institute of Clinical Physiology, CNR - Pisa - Italy, 3 Garibaldi Hospital - Catania - Italy, 4 P.O.Giovanni Paolo II - Lamezia Terme - Italy, 5 Ospedale Civico - Palermo - Italy, 6 Sandro Pertini Hospital - Rome - Italy, 7 OSP. SS. Annunziata ASL Taranto - Taranto - Italy, 8 Centro di Ricerca e Formazione ad Alta Tecnologia nelle Scienze Biomediche - "Giovanni Paolo II" - Campobasso - Italy, 9 San Francesco Hospital - Nuoro - Italy, 10 Azienda Ospedaliera Civile - O.M.P.A. Ragusa - Ragusa - Italy

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Methods. We considered 708 ß-TM patients (373 females, 30.05±9.47 years), consecutively enrolled in the Myocardial Iron Overload in Thalassemia (MIOT) network. Data were collected from birth to the first CMR imaging scan. Myocardial iron overload was assessed by the multislice multiecho T2* technique. Biventricular function parameters were quantified by cine images. Late gadolinium enhancement (LGE) images were acquired to detect myocardial fibrosis.

Results. On the basis of the type of gene mutation, three groups of patients were identified: homozygotes ß+ (N=158), compound heterozygotes ß+/ß° (N=298) and homozygotes ß° (N=252). Table 1 shows the effect of genotype group on the development of different cardiac outcomes. Compared to the milder genotype group homozygotes ß+, the other two groups showed a significantly higher risk of myocardial iron overload (MIO) and left ventricular dysfunction. We recorded 90 (13.0 %) cardiac events: 46 heart failures (HF), 38 arrhythmias (33 supraventricular, 3 ventricular and 2 hypoinetic) and 6 pulmonary hypertensions (PH). No prospective association was detected between genotype group and HF and PH. The homozygous ß° group showed a significantly higher risk of arrhythmias than the homozygous ß+ group and at the limit of significance than the compound heterozygotes. Globally, homozygotes ß° showed a significantly higher risk of CC than homozygotes ß+.

Conclusion. Different genotypic groups predict the development of MIO, left ventricular dysfunction, arrhythmias and CC in ß-TM patients. These data support the knowledge of the different genotypic groups in the clinical management of ß-TM patients.