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CARDIAC INVOLVEMENT IN MUCOPOLYSACCHARIDOSES – LONG TERM FOLLOW-UP IN 39 PATIENTS.

V. Fesslova[1], R. Parini[2], A. Grassi[2], A. Rovelli[3], S. Mannarino[1], P. Salice[1], S. Ghiglia[1], F. Furlan[2], F. Brambillasca[2], M. Gioventu[1], **F. Menni**[1]

[1]Department of Cardiology and [2]Clinical Genetics of Infancy, Department of Paediatrics, Istituti Clinici Di Perfezionamento, Milano Italy [3]Bone Marrow Transplant Unit, Department of Paediatrics, Ospedale S.Gerardo, Monza, Italy

Objectives: evaluating cardiac involvement in patients (pts) with mucopolysaccharidoses (MPS) at long-term follow-up. Patients and methods: 39 pts (16 MPS I, 5 MPS II, 9 MPS III and 9 MPS IV), 19M and 20 F, age at diagnosis 8mo-11yrs (median 2,25 yrs), underwent basal cardiological and echocardiographic examination and were followed-up for 6m-17yrs (median 9yrs). Results: Cardiac involvement was found in 79.4% pts (27/39): it occurred in 10/12(83%) MPS I Hurler pts, median age 18 mo: 2-isolated mitral insufficiency (MI), 4-MI+asymmetric septal hypertrophy (ASH), 4- MI+ aortic insufficiency (AI); in 3/ 4 MPS I Scheie pts, at 7-17 yrs: MI+AI; in 5/5 MPS II pts, median age 4yrs : 1 – MI, 4-MI+AI (1 with associated ASH); in 2/10 pts (20%) MPS III pts, at 3 yrs: 1 MI, 1 MI+AI; in 6/9 (67%) MPS IV pts, median age 2.6yrs: 3 MI, 2 AI+ASH, 1 MI+AI. Seven pts with MPS I Hurler underwent haematopoietic stem cell transplantation (HSCT); 1 died early after HSCT, 1 improved and the others remained unchanged, after 2-2.5 yrs. Two other Hurler pts died – 1 of non cardiac causes and 1 in multi-organ failure. Seven of the 27 pts with heart involvement required cardiac therapy, 1 (MPS II) had mitral and aortic valve replacement at 13 yrs. Conclusions: Cardiac involvement was more relevant in pts with MPS I and II and was progressive at follow-up; cardiac state remained mostly stable after the HSCT. Pts with MPS III and IV had milder cardiac lesions occurring later