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A Prospective Observational Cross-sectional Study on the Prevalence of Congenital Heart Malformations associated to Hirschsprung's Disease.

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Objectives: to define the prevalence of associated congenital heart malformations (CHM) in patients with Hirschsprung's disease (HSCR) and to subsequently implement a personalized diagnostic algorithm.
Background: associated CHM have been detected in 5-7% of HSCR patients according to literature and, among them, septation and conotruncal development defects appeared the most frequent.
Method: all HSCR patients admitted between January 2009 and December 2013 were included in this prospective observational cross-sectional study. Cardiovascular screening included medical history, physical examination, a twelve lead electrocardiogram and an echocardiogram. Cardiac anatomy was routinely assessed by a segmental approach. Echocardiographic measurements respectively of the left ventricular dimensions and wall thickness, of the aortic root and of the left ventricular systolic and diastolic functions were obtained. CHM requiring a percutaneous or surgical intervention was described as major CHM
Results: 133 consecutive HSCR underwent the cardiac screening. Mean age at enrolment was 5,3 ± 6,1 years. Eleven patients (8,3%) presented an associated CHD (Table 1). All patients who underwent an intervention are asymptomatic and without any residual cardiac defects at a mean follow up of 9 ± 5,8 years. We observed mild dilatation of aortic root in 3 patients whereas in other 2 measurements were within the superior normal limits.
Conclusions: in our series the prevalence of associated CHMs was slightly higher than in the previous papers, and were mostly represented by septal defects. Noteworthy, no one presented conotruncal heart defects. Six patients (4,5%) had major CHM and 4 of them had chromosomal abnormalities. If we do not consider the subpopulation of patients with HSCR and associated chromosomal anomalies, cardiac defects were still present in approximately 3.8% of the patients, which means that the prevalence of CHM in isolated HSCR population is higher than that of the general population. Basing on these results we suggest to perform routine echocardiogram in all HSCR patients, with or without associated chromosomal syndromes.

Patient	Sex	Age	Type of Heart Disease	Management	Syndrome
1	M	59	OS ASD + small PDA	F-up	
2	M	58	OS ASD + small PM VSD	F-up	Down
3	F	54	OS ASD	F-up	
4	M	51	OS ASD + moderate PM VSD	Intervention	
5	F	309	OP ASD	Intervention	Down
6	M	171	OS ASD + moderate PM VSD	Intervention	Down
7	M	192	large PM VSD	Intervention	Down
8	F	27	CoA	Intervention	Turner
9	M	96	OS ASD	Intervention	
10	M	52	tiny PDA	F-up	Down
11	M	107	small PDA	F-up	