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Males with 45,X/46,XY have similar cardiovascular problems as females with Turner syndrome

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Cardiovascular pathology in Turner syndrome is well described and afflicts 25 to 45% of patients. We hypothesised that males with mixed gonadal dysgenesis and 45,X/46,XY (MGD) have similar cardiovascular problems as female Turner patients with 45,X/46,XY (TS).

Patients and methods: in a multicenter Belgian study, we collected 16 patients with mosaicism 45,X/46,XY who were followed at an endocrinology department: 6 females with Turner syndrome and 10 males with mixed gonadal dysgenesis. Age ranged from 1 to 38 y with a median of 13 y. Data on sexual phenotype and growth were collected from the files. Patients underwent blood pressure measurement, ECG, echocardiography and MRI (in 10/16).

Results: Turner patients with 45,X/46,XY (age 13-38 y) had normal female genitals, short height and delayed pubertal induction. All received growth hormone and hormone replacement therapy. Males with 45,X/46,XY (age 1-24y) had abnormal genitals ranging from minor abnormality (EMS12) to ambiguous genitals (EMS5). Seven received growth hormone, the other 3 were still too young.

All patients had normal blood pressure. Prolonged QTc was found in 5 (3/10 MGD, 2/6 TS). Structural heart abnormalities were equally found in TS (3/6) and MGD (5/10). Males with MGD had a high incidence of bicuspid aortic valve (50%) and dilation of the ascending aorta (20%). Both cases of dilated ascending aorta were in young boys (age 10 y) who were not followed at the cardiology department. Dilation was important (28 and 30 mm). Both boys also had a bicuspid aortic valve and one had an impaired ventricular function.

Structural cardiac abnormality	MGD (n=10)	TS (N=6)
VSD	1	0
Bicuspid aortic valve	5	2
Dilation of the ascending aorta	2	0
Coarctation	0	1
Right arteria lusoria	0	1

Conclusion: males with 45,X/46,XY have the same cardiovascular defects as females with Turner syndrome. Dilation of the ascending aorta can be important and might become life threatening. We advise a cardiac screening in all males with 45,X/46,XY. As in Turner syndrome, life-long follow up may be recommended.