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Original Research Paper

Medicine

A CASE REPORT OF MARFAN SYNDROME WITH ISCHEMIC STROKE WITH AORTIC DISSECTION

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ABSTRACT

Marfan syndrome is an autosomal dominant disorder of connective tissue with musculo-skeletal, ocular and cardiovascular manifestations. Mutations in the gene encoding fibrillin on chromosome 15 constitute the likely underlying cause in majority of cases. Clinical expression of genetic defect, however can be variable both within and between families. In large and medium sized arteries, defects in fibrillin are associated with the disruption of elastic fibers, predisposing to aneurysm formation and arterial dissection. These vascular abnormalities can be a cause of cerebral or spinal ischemia or hemorrhage. Indeed ischemic events involving brain or spinal cord are estimated to occur in 10 to 20% of patients with Marfan syndrome.Aortic dissection is a major contributor to the premature mortality of Marfan syndrome. Extension of dissecting aortic aneurysm into the brachiocephalic and common carotid arteries may lead to ischemic stroke. Knowledge of the vascular complications of Marfan syndrome has come mainly from case reports involving small number of patients with concomitant potential for selection bias. Henceforth, here we report a case of Marfan syndrome who presented with ischemic stroke and aortic dissection.

KEYWORDS :

CASE REPORT:

HISTORY:

A 34 year old male coming from Anand from lower socioeconomic status presented to us in OPD with chief complaint of right sided hemiparesis since 1 day.

GENERAL EXAMINATION:

He was well oriented to time, place and person with temperature and pulse rate within normal limits and his blood pressure was 140/90 mm of Hg. He was tall and thin with increased arm span/height ratio. Upper segment/lower segment ratio is lower with significant facial features like dolicocephaly and malar hypoplasia. Wrist and thumb sign was positive and pectus carinatum was found on examination of thorax. Similar features were also present in his father and sister on their physical examination.

CVS EXAMINATION:

Early diastolic murmur present at left 3rd intercostal space best heard at end-expiration phase.

CNS EXAMINATION:

Decreased tone on right side with extensor plantar and exaggerated reflexes on right side with rest of the examination being normal.

INVESTIGATIONS:

CBC, RFT and LFT were within normal limits. ECG showed features of left axis deviation with left ventricular hypertrophy. 2D Echo showed ejection fraction to be 35% with global LV dysfunction and moderate MR and moderate AR with flap like structure extending upto ascending aorta.

Chest xray showed congestive changes in both the lung fields with cardiomegaly. MSCT brain showed hypodensity in left insular cortex and left gangliocapsular region with hyper dense middle cerebral artery s/o acute infarct. MSCT angiography aorta showed dilated aortic root and ascending aorta with Stanford Type A dissecting aneurysm.

Ophthalmic examination showed myopia and subluxation of lens in left eye to left upper outer quadrant.

CLINICAL COURSE AND TREATMENT:

Patient was started on injection mannitol and Lasix with tablet aldectone, warfarin, aspirin, atorvastatin, metoprolol and Ramipril. Surgical options for aortic root replacement:

(1) Composite graft valve: Bentall procedure

(2) Valve sparing aortic root replacement: David reimplant

DISCUSSION:

Marfan syndrome is a multisystem connective tissue disorder usually associated with mutation in fibrillin, and occasionally with mutation in TGFBR1 or 2. The clinical diagnosis is made using the Ghent nosology, which will unequivocally diagnose or exclude Marfan syndrome in 86% of cases. Use of a care pathway can help implementation of the nosology in the clinic. The penetrance of some features is age dependent, so the nosology must be used with caution in children. Molecular testing may be helpful in this context. The nosology cannot be used in families with isolated aortic dissection, or with related conditions such as Loeys–Dietz syndrome, although it may help identify families for further diagnostic evaluation because they do not fulfill the nosology, despite a history of aneurysm. Prophylactic medical (eg b-blockade) and surgical intervention is important in reducing the cardiovascular complications of Marfan syndrome. Musculoskeletal symptoms are common, although the pathophysiology is less clear – for example, the correlation between dural ectasia and back pain is uncertain. Symptoms in other systems require specialist review such as ophthalmology assessment of refractive errors and ectopia lentis. Pregnancy is a time of

increased cardiovascular risk for women with Marfan syndrome, particularly if the aortic root exceeds 4 cm at the start of pregnancy. High-intensity static exercise should be discouraged although low-moderate intensity dynamic exercise may be beneficial. The diagnosis and management of Marfan syndrome requires a multidisciplinary team approach, in view of its multisystem effects and phenotypic variability.

Table 2. Main diagnostic work-up for Marfan's syndrome

Work-up	Target
Echocardiogram, aortogram, magnetic resonance imaging and computed tomography	Measurement of the aortic root and detection of valve prolapse
Slit lamp examination	Lens abnormalities
X-ray studies on skeletal system	Evaluations of hand, spine, pelvis, chest, foot and skull for characteristic abnormalities
Magnetic resonance imaging	Dural ectasia
Prenatal testing	At approximately 10-12 weeks, using chorionic villus sampling, on a prospective parent who has Marfan syndrome
Genetic testing	Genetic testing may be helpful, but is very costly and time-consuming for different gene mutations

P		Systemic involvement (>7 point)
3	OR	Wrist AND thumb sign
1		Wrist OR thumb sign
2	OR	Pectus carinatum
1		Pectus excavatum OR chest asymmetry
2	OR	Hindfoot deformity
1		Plain pes planus
2		Pneumothorax
2		Dural ectasia
2		Protusio acetabuli
1		USLS‡ AND ASHR† AND no scoliosis
1		Scoliosis OR thoracolumbar kyphosis
1		Reduced elbow extension
1		Facial abnormalitis (see above)
1		Striae atrophicae
1		Myopia > 3 diopters
1		Mitral valve prolapse

Diagnostic scenarios	
I	Aortic involvement AND Ectopia lentis
II	Aortic involvement AND FBN1 mutation
III	Aortic involvement AND Systemic involvement
IV	Aortic involvement AND Family history
V	Ectopia lentis AND FBN1 mutation
VI	Ectopia lentis AND Family history
VII	Systemic involvement AND Family history

Revised Ghent nosology (P: systemic score point; ASHR: arm span to height ratio, USLS: upper segment to lower segment ratio, FBN1: fibrillin-1 gene).

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