Case reports

A child with Morquio syndrome and mixed mitral valve disease
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Introduction

Mucopolysaccharidoses (MPSs) are a group of inherited storage diseases caused by a deficiency of lysosomal enzymes that degrade glycosaminoglycans (GAGs). The MPSs are a heterogenous group characterised by intra-lysosomal accumulation of GAGs, excessive urinary excretion of GAGs, and variable degrees of progressive mental and physical deterioration [1]. Mucopolysaccharidosis type IV (Morquio syndrome) is autosomal recessively inherited. The primary pathology is defective degradation of keratan sulphate due to one of two enzyme deficiencies; n-acetylgalactoseamine-6-sulfate (chromosome 16q) or β-galactosidase (chromosome 3p) [1,2].

Case report

A 10-year old child was admitted to hospital with a history of fever, cough and exertional dyspnoea for 5 days. She was clinically diagnosed to have Morquio syndrome at the age of 2 years and had short stature, skeletal dysplasia, corneal clouding and normal intelligence. She gave a history of frequent episodes of shortness of breath during the past 2 years.

On examination her height and weight were both well below the third centile. She had a short trunk, pectus carinatum, corneal clouding, and short stubby hands and fingers. She was tachypnoeic, and using accessory muscles of respiration. On auscultation bilateral basal crackles were heard. She had also tachycardia and cardiomegaly. There was a pansystolic murmur and a diastolic murmur best heard at the apex. Liver was palpable 3 cm below the right costal margin and the spleen was just palpable. Neurological examination was normal except for a waddling gait.

Chest xray showed cardiomegaly, double and straight left heart border and plethoric lung fields (figure). ECG showed bifid P waves (P mitrale). 2-D echocardiogram showed a dilated left atrium, moderate mitral stenosis, grade III mitral regurgitation and mild tricuspid regurgitation.

Discussion

Characteristic features of Morquio syndrome are short-trunk dwarfism, skeletal dysplasia (including thoraco-lumbar kyphosis, atlanto-axial subluxation, anterior vertebral beaking, pectus carinatum, rib abnormalities, coxa valga, genu valgum, epiphyseal/metaphyseal irregularities and small irregular metacarpals, corneal clouding, and waddling gait with preservation of intelligence [1], 2].

Cardiac involvement is well recognised in most forms of mucopolysaccharidoses but there is poor documentation of abnormalities specific to Morquio syndrome. Cardiac involvement is usually of regurgitant mitral and aortic valve lesions due to cardiomyopathy [2]. In a study of 10 patients of Morquio syndrome, 5 patients had mitral valve involvement and 4 had aortic valve involvement. Only a single patient had mitral valve thickening to the point of mitral stenosis. All cardiac lesions were mild and haemodynamically stable [3]. A search of the literature revealed only this one reported case of mitral stenosis in a child with Morquio syndrome.

The index patient had clinical, radiological, and echocardiographical evidence of moderate mitral stenosis and moderate to severe haemodynamic instability. This is the second reported case of mitral stenosis in a child with Morquio syndrome and in contrast to the previous case, this child has severe mitral valve disease with haemodynamic instability. She was started on medical treatment with diuretics and calcium channel blockers and is awaiting mitral valve replacement.

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A case of generalised myasthenia gravis with membranous nephropathy

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Abstract

We report a 40-year old woman with bilateral partial ptosis, complete external ophthalmoplegia, and weakness and fatiguability of upper limbs. She was on treatment for hypertension for 5 months at the time of admission. She was found to have generalised myasthenia gravis and membranous nephropathy with end-stage renal disease. Her symptoms and signs improved within 2 months on treatment with neostigmine and prednisolone. It is postulated that either thymic hyperplasia or the subclinical stage of a thymoma may be the underlying aetiological factor in this patient.

Introduction

Myasthenia gravis is an autoimmune syndrome that is rarely associated with glomerulonephritis [1, 2]. The types of glomerulonephritis described in association with myasthenia gravis include minimal change disease, focal segmental glomerulosclerosis and membranous nephropathy [1, 3, 4]. We report here a patient with generalised myasthenia gravis and membranous nephropathy.

Case report

A 40-year old Sri Lankan woman with hypertension for 5 months, presented with a 6 weeks' history of involuntary closure of both eyes. Examination revealed bilateral partial ptosis and bilateral complete external ophthalmoplegia. There was fatiguability of both eye lids. Muscle power was reduced to grade IV in abductors and extensors of the right arm. All tendon reflexes were normal. Blood pressure was elevated.

The diagnosis of myasthenia gravis was confirmed by a positive edrophonium test and a decremental response of 23% in facial muscles by the repetitive nerve stimulation test. Acetylcholine receptor antibody assay and single fibre electromyogram were not available locally. Urine analysis revealed a urinary protein excretion of 4.3g/day. Creatinine clearance was 12.7 ml/ per minute. There was hypoalbuminaemia and hypercholesterolaemia. Renal ultrasound scan revealed normal sized kidneys with increased cortical echogenicity. Renal biopsy established membranous nephropathy (figure 1). Extensive glomerular sclerosis and capillaries with spiking of the basement membrane, extensive lymphocytic interstitial inflammation and fibrosis are evident. Immunofluorescent staining was positive for IgG and negative for IgA, IgM and C3. Chest xray and CT scan of the head and orbits were normal. CT scan of the thorax showed a normal sized thymic hyperdensity (figure 2). The ESR was 60mm/hour. Antinuclear antibodies, rheumatoid factor, and hepatitis B, hepatitis C, VDRL and HIV antibodies were negative. Screening for malignancy with stools for occult blood, CT abdomen, carcinoembryonic antigen and CA 125 were negative.

Ptosis and external ophthalmoplegia showed complete improvement after about 2 months of neostigmine and prednisolone. The patient is now completely free of myasthenia gravis symptoms on a prednisolone dose of 7.5mg daily. She is being prepared for renal transplantation.

Discussion

Myasthenia gravis with membranous nephropathy is a rare association [1, 4, 5, 6]. Their observations tally with the natural history of membranous nephropathy. In contrast our patient was already in end-stage kidney disease at the time of diagnosis. Two large case series comprising of 206 myasthenia gravis patients have failed to detect any with the combination of myasthenia gravis and membranous nephropathy.