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Case Series

Sinoatrial Node Disease in Adults with Down's Syndrome.

Kennedy J, Devlin P, Wilson CM, McGlinchey PG

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INTRODUCTION

Down's syndrome, also known as trisomy 21, is a common chromosomal abnormality associated with multiple co-morbidities, premature ageing and decreased life expectancy. We present four cases of individuals with Down's syndrome without congenital heart disease who presented with syncope and subsequently were found to have severe sinoatrial node disease, all requiring permanent pacemaker implantation. An association between the conditions has not previously been described and we postulate a possible increased frequency of sinoatrial node disease in adults with Down's syndrome. In addition, in each case there was a considerable delay in reaching the diagnosis of sinoatrial node disease; this was partly due to alternative differential diagnoses (particularly neurological) being considered first, partly due to difficulties in obtaining an accurate history of the syncopal events and partly due to decreased co-operation by the patients with regards to investigation of the underlying aetiology of their events.

CASE SERIES

Case 1. A 70 year old man with Down's syndrome was admitted following two syncopal events. He was placed on cardiac monitoring and had a number of episodes of sinus arrest of up to fifteen seconds. Nine months earlier, he had been admitted with a syncopal episode to another institution, where he had been uncooperative with medical staff, refusing all examination and had been provisionally diagnosed with a transient ischaemic attack. A permanent pacemaker was implanted on this occasion under general anaesthesia. Although he had no further syncopal episodes after implantation, he has subsequently died around a year later from an unrelated illness.

Case 2. A 54 year old man with Down's syndrome was referred by his General Practitioner following two episodes of syncope. He had been found lying on the ground. There was no seizure activity and he was noted to be pale. There was no tongue biting or urinary incontinence. He was assessed in an Emergency Department, with no abnormality found. He was referred to a neurologist in the first instance and the possibility of atonic seizures was raised and further investigation planned.

Resting ECG showed sinus rhythm with first degree atrioventricular block and incomplete right bundle branch block. Carotid sinus massage showed physiological slowing of heart rate only. Holter monitoring showed sinus rhythm, heart rate varying between 51 and 95 beats per minute. There were rare atrial and isolated ventricular ectopic beats but no arrhythmia was found to explain his presentation.

One year later, he presented as an emergency admission with four further episodes of syncope over a 48 hour period. He was placed on cardiac monitoring which demonstrated sinus pauses of nine and fourteen seconds. He subsequently had a permanent pacemaker implanted under general anaesthesia. He has not had any further syncopal events.

Case 3. A 56 year old man with Down's syndrome was referred with frequent syncopal events. He lived in residential accommodation and when staff noticed these events he was found to be very bradycardic. He refused to comply with wearing a Holter monitor or having an ECG performed. His radial pulse was 40 beats per minute by palpation. His case was discussed with a pacemaker implanter who felt that there was not enough evidence to support permanent pacemaker implantation.

Seven months later, he presented as an emergency with recurrent syncopal events. Sinus pauses of up to nine seconds were recorded. He has had a permanent pacemaker implanted under general anaesthesia with no further syncopal episodes to date.

Case 4. A 46 year old man with Down's syndrome was referred following assessment by a neurologist, who in turn had been referred the patient by an acute physician. There was a one year history of episodes of sudden collapse associated with pallor. There was a background history of epilepsy but the more recent episodes were felt to be different. A provisional diagnosis of atonic seizures had been made but the neurologist was not convinced. There were no abnormalities of 12-lead ECG and carotid sinus massage was normal. A patient activated monitor was fitted and when the patient was syncopal, sinus pauses of up to 15 seconds were documented. He subsequently has had a permanent pacemaker implanted under general anaesthesia and remains well.

Department of Cardiology, Altnagelvin Area Hospital, Glenshane Road, Derry, Northern Ireland, UK BT47 6SB

paul.mcglinchey@westerntrust.hscni.net

Correspondence to Dr Paul McGlinchey

CONCLUSION

Down's syndrome, is a common genetic disorder with an incidence of around 0.1% of live births[1]. Individuals with Down syndrome have a higher risk of early death than the general population[2]. Following improved medical care life expectancy has increased[1]. About 10% of individuals with Down's syndrome but without congenital heart disease live to 70 years of age[3], the implication of which is that we have a greater prevalence of older individuals with Down's syndrome than in the past.

We present four cases of adult males with Down's syndrome, but without history of congenital heart disease, who have presented with syncope due to severe sinoatrial node disease requiring permanent pacemaker implantation. This raises the possibility of an association between the two conditions that has not been previously reported. We postulate that improvements in life expectancy in individuals with Down's syndrome will lead to an increased prevalence of patients with this condition presenting with sinoatrial node disease; further epidemiological data will be needed to confirm or refute this postulation.

In each case, there was a delay in making the diagnosis of underlying sinoatrial node disease. This was due to initial presentation and/or referral to specialties outside of cardiology, most frequently a provisional diagnosis of a neurological condition had been made. This was compounded by difficulties in obtaining an accurate history of the events and poor co-operation by the individual patients in regards to investigation. We suggest that underlying conduction tissue disease, most likely of the sinoatrial node, should be high on the list of differential diagnoses when considering unexplained syncope in adults with Down's syndrome.

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