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OUTPATIENT MEDICAL REPORT

Ani Mihailova – Dob: 18/12/2007

DIAGNOSIS

Possible genetic aortopathy

HISTORY

I was delighted to see this five year old girl from Bulgaria for cardiac evaluation on the 20th August at the request of my colleague Dr. Saggar, Consultant Paediatric Geneticist. Ani has been under investigation for possible Marfan's syndrome. She developed eye problems at the age of eight months but did not require surgery. She also has scoliosis and is double-jointed with general floppiness. She was found to have hypothyroidism at the age of twenty-one days during a screening programme and is taking thyroid replacement.

Ani was seen for cardiac review earlier this year and mitral valve prolapse was diagnosed. She has been generally well and has had no other symptoms. Her mother has mitral valve prolapse and supraventricular arrhythmia. She has one brother who is 1 m, 90 cm tall but is well. There is no family history of cardiac disease or Marfan's syndrome.

EXAMINATION

Not typical Marfan's. Normal peripheral pulses. No clinical evidence of cardiac failure. Quiet precordium. Auscultation revealed normal heart sounds and no murmurs.

MANAGEMENT

I performed an ECG which showed sinus rhythm with a heart rate of 93 bpm and a PR interval of 140 ms. QT interval was normal and there was no evidence of pre-excitation or chamber enlargement.

Cross sectional echocardiography showed a structurally normal heart, Left ventricular function was normal (ejection fraction of 68%). There was no evidence of mitral valve prolapse or regurgitation (mitral valve diameter 22 mm). There was mild tricuspid regurgitation with very mild tricuspid valve

prolapse (diameter 22 mm). Right ventricular pressure was normal. Appearances of the aortic root were normal (aortic valve diameter 15 mm, aortic sinus diameter 1 mm, ascending aorta diameter 18 mm, transverse aortic arch diameter 13 – 14 mm, descending aorta diameter 11 mm). Doppler flows throughout the heart and great arteries were normal. There was no evidence of a structural heart defect.

Ani does not have typical features of Marfan's syndrome. She does however very likely have a genetic aortopathy, which is under investigation by Dr. Saggar. She will need to stay under regular cardiac review and should be seen at six months – one yearly intervals. I would be delighted to take part in Ani's cardiac follow up and review any investigations undertaken in Bulgaria. Alternatively, I would be very pleased to see the family when they next come to London.

Thank you for referring this super patient to me.

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