

A Marfanoid Syndrome in a School Girl Revealed by an Ectopia Lentis

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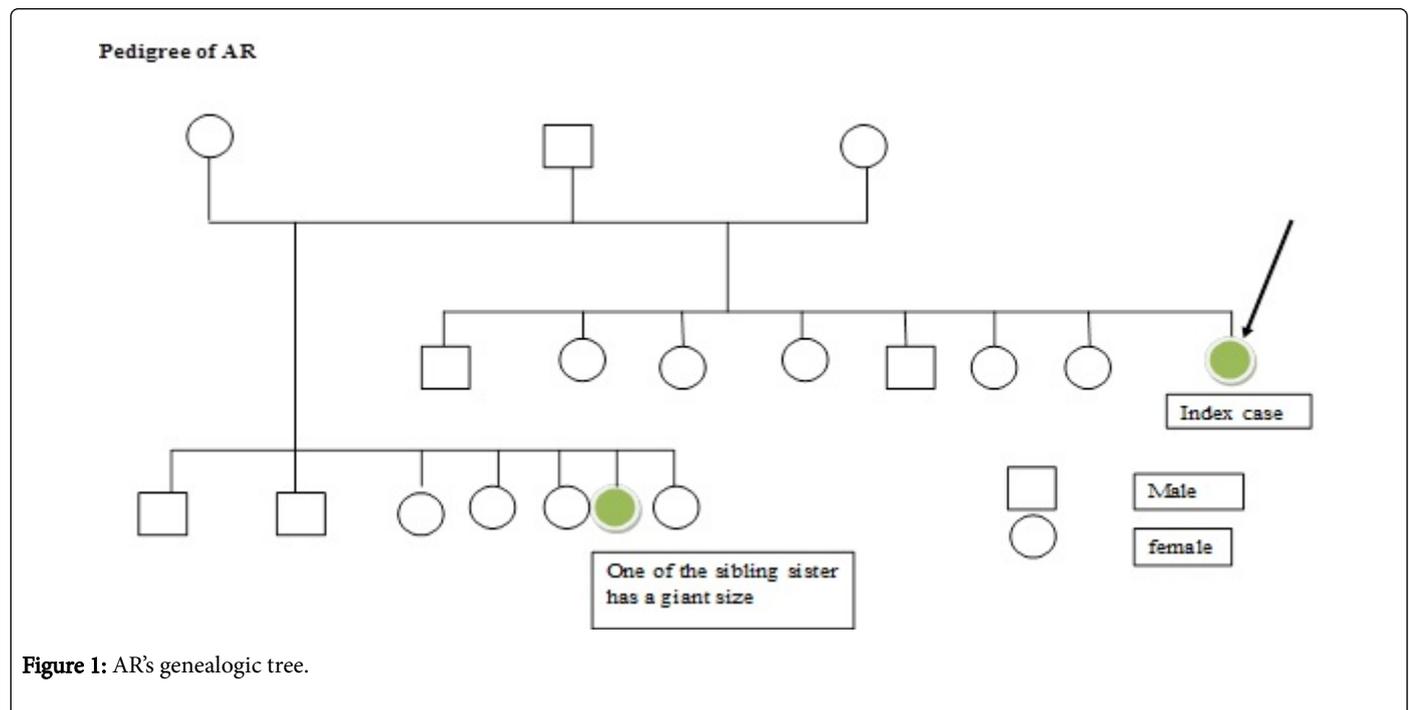
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Clinical Observation



Presenting complaint

R.A was brought by her senior sister for pediatric examination/visit due to suspicion of Marfan syndrome on April 9, 2014.

Anamnesis/History of the presenting complaint

R.A. is a little girl of 8 and a half years brought by her senior sister for pediatric medical examination due to suspicion of Marfan Syndrome at the request of a Senior ophtalmologist of Lome (Rep. of Togo) further to an ectopia lentis [1,2] discovered after an ophthalmologic exam for visual impairment two days before i.e. April 7, 2014.

The pathology dates back many years and started with a visual impairment in the class room where the little girl put a lot of effort to read on the chalkboard. After several exams in different eye clinics and other health centers and on the decision of the child's father, an ophthalmologic exam performed in Lome helped find out an ectopia lentis.



Figure 2: Positive thumb sign.

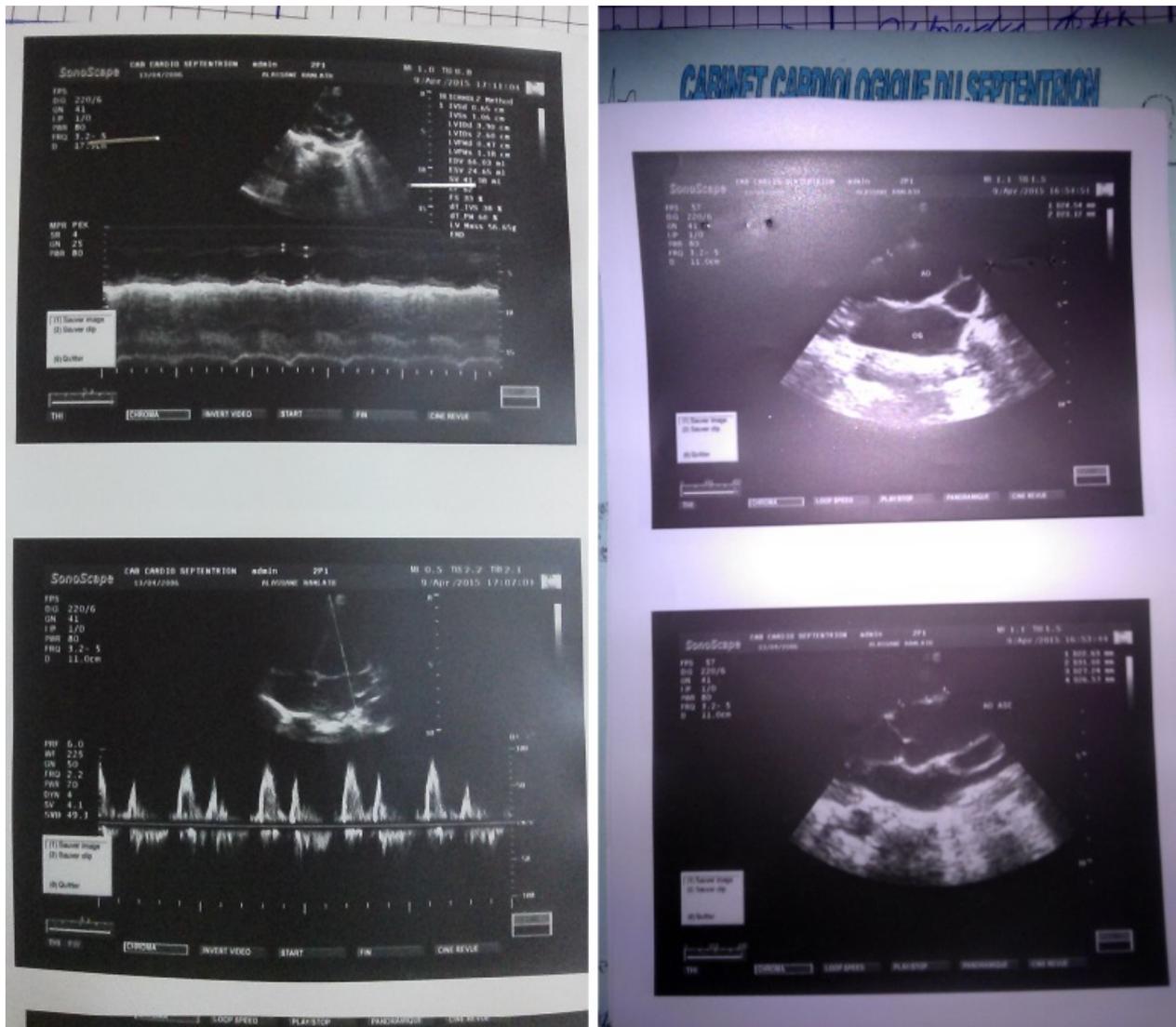


Figure 3: Normal cardiac ultrasonography.

Health history

She is the 8th child of 8 maternal siblings. As a whole, the siblings were 15 children (Figure 1).

Developmental history

She is in Grade 3 of primary school with good academic records. Her psychomotor development was characterized by a delay in walking at the age of 2 years but language was acquired at 18-24 months of age.

Pregnancy and delivery history

Pregnancy and childbirth were well conducted. The senior sister didn't remember any special perinatal pathology while A.R. was given birth except a postpartum hemorrhage of small amount from their mother.

Immunization history and other prophylaxis

Based on Expanded Programme on Immunization, her immunization status would be up to date. The same applies to antimalarial prophylaxis.

Feeding history

The diet would have been well managed; Notion of glaucomatous disease in the Father.

Family history (Inherited diseases)

Notion of large size among siblings and particularly in one of the half-sisters on the father's side.

Examination

Weight 30 kg, large size of 1.53 m (normal size \approx 1.08-1.12 m), well above the 97th percentile of Sempe curve (WHO universal charts stop

at 5 years), temperature: 36.1°C, respiratory rate: 32 cycles/mn, heart rate: 70 cycles/mn, Full arm span: 1.57 cm, Arm span/ size Ratio = 1.02 (VN < 1.05).

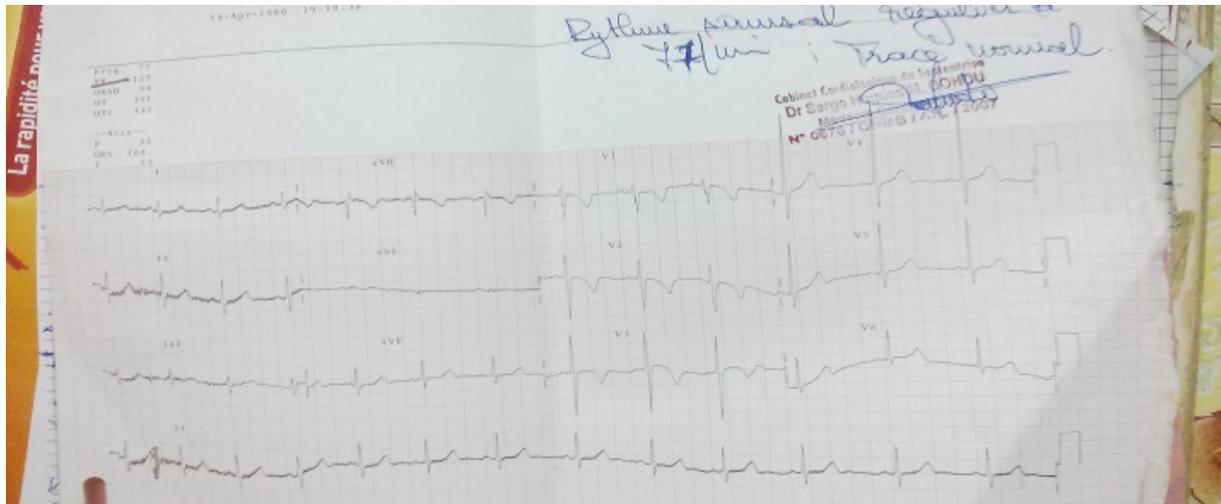


Figure 4: Normal ECG AR's.

Positive thumb sign (Figure 2). There was no scoliosis, no joint hypermobility; elbow extension is normal at 180° (abnormal if < 170°).

The rest of clinical exam seems normal. A cardiology consultation was requested. ECG was normal and Echo Doppler heart test does not detect abnormality in the ascending aorta though AR clinically presents Marfanoid features, without cardiac signs (Figures 3 and 4).

Suspected diagnosis

AR a little girl of 8½ years have been examined for developing signs of Marfanoid syndrome, probably partial or incomplete in terms of

large size, ectopia lentis, positive thumb sign and possible family predisposition [1,2].

References

1. Stheneur C, Chevalier B. Syndrome de Marfan (2008) In *Pédiatrie pour le praticien*. Bourillon 209-10. 6th Edition Elsevier Masson, Paris 2008.
2. Loeys BL, Dietz HC, Braverman AC, Callewaert BL, Backer JD, et al. (2010) The revised Ghent nosology for the Marfan Syndrome. *J Med Gen* 47: 476-485.