Case Report: Dextrocardia and PHACES Syndrome

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Case History

41-year-old Caucasian female, Uniformed Services Scientist, presented to Family Medicine Practice to establish primary care. She requested her yearly physical exam along with an ECG to monitor her dextrocardia.

The patient had a past medical history of dextrocardia, enlarged segmental facial hemangioma as a child s/p resection, right ear hearing loss (25dB) affected by the hemangioma, ventral septal defect, murmur, migraines, H/O amblyopia with Lasix vision correction surgery, scoliosis, spina bifida occulta, teratoma with left oophorectomy, pericallosal lipoma and small lipomas in the choroid plexus of the lateral ventricle, asymmetric thyroid lobes, anatomical variations to the circle of Willis and the neck vessels, insomnia, increased BMI, high blood pressure without a diagnosis of hypertension, hyperlipidemia, and panic disorder.

The patient was diagnosed with dextrocardia as a child and was followed by Pediatric Cardiology until the age of 14. At the time of visit, the patient had no acute cardiac complaints. She had a history of abnormal ECGs, consistent with dextrocardia.

Her cardiac Review of Systems was negative for chest pain and palpitations.

Family History

Family history of hypertension, heart disease, and diabetes.

Physical Examination

Vitals: BP: 122/84, pulse: 80, SpO_2: 99%, BMI: 28.89 kg/m^2.

Constitutional: Normal healthy/athletic appearance.

HEENT: Head is normocephalic and atraumatic. Extraocular movements intact.
Residual telangiectatic areas of lower lip and right ear.
Midline lower lip scar crossing of the vermilion border.

Cardiovascular: Normal rate and regular rhythm. Loudest along right sternal border.

Pulmonary: Effort is normal. Normal breath sounds.

MSK: Normal range of motion.

Neurological: No focal deficit present. Alert and oriented to person, place, and time.

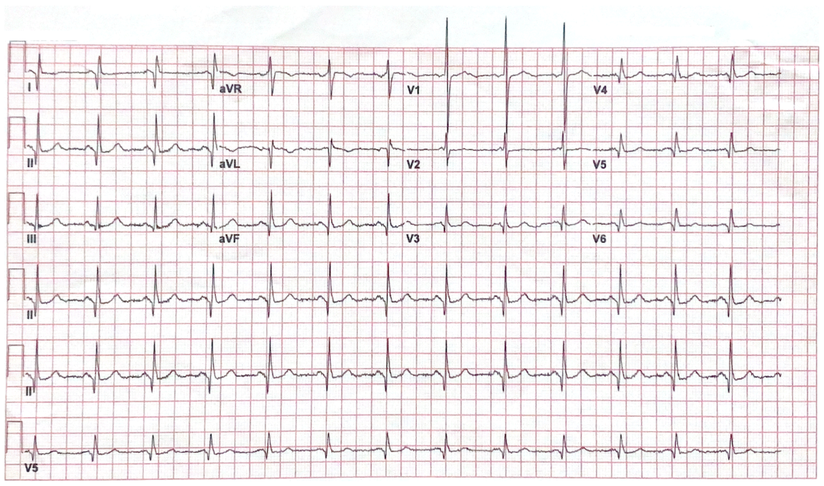
Psychiatric: Mood normal. Behavior normal.

Previous ECHO findings

Transthoracic echocardiogram:
*Atrial situs solitus
*Atrioventricular concordance
*d-looped ventricular topology
*Pulmonary artery left and anterior to aorta
*Normal chamber sizes
*Normal right ventricular and left ventricular systolic performance
*Normal valve function

ECG

Ventricular rate: 78 bpm
PR interval: 134 ms
QRS duration: 90 ms
QT/QTc: 372/424 ms
PRT axes: 63/88/87
NORMAL sinus rhythm
Lateral infarct, age undetermined



Tests & Results

TSH 1.71 (0.27-4.20)
HA1C 4.9% (<5.7%)
CMP WNL other than ALT
ALT 44 (<41)
Total Cholesterol 182 (<200)
Triglyceride 232 (<150)
HDL 56 (>60)
LDL 87 (<100)
Non-HDL 125 (<130)
ASCVD Risk 0.4%
Uric Acid 6.5 (2.4-5.7)
CBC with Diff WNL

MRI Brain W/O contrast: Findings consistent with a pericallosal lipoma, as well as small lipomas in the choroid plexus of both lateral ventricles. Asymmetry in the thyroid lobs with the right being larger than the left. Although the left lobe appears small, the right lobe is heterogeneous. A thyroid ultrasound would be helpful to characterize these findings further.

Discussion

The patient's dextrocardia is part of a larger and extremely rare multifactorial syndrome called PHACES. Dextrocardia situs inversus totalis has an incidence of .008% (1 in 12,000).[1] As of 2022, the incidence of PHACES is 4% or 5%. There has been no single genetic abnormality that causes this syndrome. Rather, it is multifactorial, assumed to be a combination of environmental trigger and genetic predisposition. It is poorly understood but categorized as a neurocutaneous disorder encompassing the following findings:

Posterior fossa malformations (ex Dandy-Walker)

cervicofacial **H**emangiomas

Arterial abnormalities

Cardiac anomalies,

Eye anomalies

Sternal clefting with and without supraumbilical raphe.[2,3]

The patient was finally diagnosed with PHACES at the age of 31, although this constellation of symptoms was present since birth.

Arterial abnormalities in PHACES can include coarctation of the aorta, aberrant subclavian artery, and ventricular septal defect (VSD), which was the arterial abnormality in this patient. Her VSD spontaneously closed.

The patient was assessed for dextrocardia and PHACES ten years prior and received a transesophageal echocardiogram and an ECG. Her ECG revealed a right-shifted apex and atrial situs solitus with AV concordance; her aortic arch was normal. She was deemed New York Heart Classification 1.

Because PHACES syndrome has a risk of long-term morbidity, systematic approaches to screening—such as determining which patients should be screened—are warranted. Garzon et al. (2016) outline a screening approach:

infants with facial Infantile Hemangioma (IH) of 22 cm^2 (5 cm x 4.5 cm)

infants with smaller IH and PHACE findings

infants without IH with PHACE findings

Screening should include physical exam, echocardiogram with possible referral to pediatric cardiologist, MRI of head and neck with possible referral to neurosurgeon if structural abnormalities are identified, MRA of head and neck to identify cerebral vascular anomalies, and an ophthalmology exam.[4]

Plan

Dextrocardia, as an isolated finding, is benign and does not require special monitoring. ECG will be updated and compared to prior ECGs. Patient will be referred to cardiology if appropriate, otherwise routine monitoring is not warranted.

Referral to Neurology if migraines persist.

Recommend routine follow up with Ophthalmology.

Recommend thyroid ultrasound for further evaluation.

Recommend further work up for elevated uric acid level and ALT.

No activity restrictions were recommended at this time.

References

1 Nair, Raunak, and Sujit R. Muthukuru. "Dextrocardia." StatPearls [Internet]. StatPearls Publishing, 2021.
2 Chamli, Amal, and Noureddine Litaiem. "PHACE Syndrome." StatPearls [Internet]. StatPearls Publishing, 2021.
3 Proisy, Maia, et al. "PHACES syndrome and associated anomalies: risk associated with small and large facial hemangiomas." *American Journal of Roentgenology* 217.2 (2021): 507-514.
4 Garzon, Maria C., et al. "PHACE syndrome: consensus-derived diagnosis and care recommendations." *The Journal of pediatrics* 178 (2016): 24-33.