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CASE REPORT

Unexpected finding of thyroid hemiagenesis in a patient presenting with a right thyroid nodule and a history of Poland syndrome

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Introduction: Poland syndrome is defined primarily by congenital absence of the pectoralis major. Thyroid hemiagenesis is the congenital absence of a single thyroid lobe. The combination of both pathologies has not been previously described.
Case presentation: Poland syndrome is a rare congenital condition characterised by aplasia or hypoplasia of the pectoralis major muscle, associated to varying degrees with malformation of the ipsilateral upper limb. It is often accompanied by other congenital abnormalities, but the presence of a single thyroid lobe is exceptional. We report to our knowledge the first observation of thyroid hemiagenesis and Poland syndrome.
Observation: A 19-year-old female was referred for chronic right neck swelling. On physical examination, she had a relatively small stature but an overall normal appearance. Neck examination revealed a mobile 3 cm right firm thyroid nodule. There was complete absence of the left breast and pectoralis muscles, with normal development on the right. The left hand was relatively small and exhibited syndactyly. Biochemical testing revealed normal thyroid function. Neck ultrasound revealed complete absence of the left thyroid lobe and the presence of a well-circumscribed hypoechoic 3 cm right thyroid nodule without calcifications, TIRADS score 3. The patient was diagnosed with Poland syndrome, a 3 cm right thyroid nodule, and agenesis of the left thyroid lobe. Her consent was obtained for a right thyroid lobectomy due to the cosmetic impact of the nodule, as well as the small risk that this represented a thyroid malignancy. An uncomplicated right thyroid lobectomy was performed. Final surgical pathology was consistent with a 3 cm benign follicular adenoma.
Conclusion: Poland syndrome and thyroid hemiagenesis are both rare congenital malformations. Their association has not been previously described. Prior to surgery, the patient must be informed of the mandatory need for lifelong thyroid hormone replacement.

Introduction

Poland syndrome is defined primarily by congenital absence of the pectoralis major.[1,2] Thyroid hemiagenesis is the congenital absence of a single thyroid lobe.[3,4] The combination of both pathologies has not been previously published. We report to our knowledge the first observation of thyroid hemiagenesis and Poland syndrome, in a young Senegalese female.

Case presentation

A 19-year-old female was referred to the otolaryngology department of a major tertiary referral and teaching hospital in Dakar, Senegal, for chronic right neck swelling, now causing a degree of physical disfigurement (Figure 1). She denied any compressive symptoms, such as pain, pressure, dysphagia, dyspnoea or voice change. She denied systemic symptoms of fever, sweats or weight loss. There were no symptoms of hypothyroidism or hyperthyroidism.

On physical examination, she had a relatively small stature but an overall normal appearance. There were no overt signs of hypothyroidism or hyperthyroidism. Vitals were within normal limits, without tachycardia. Neck examination revealed a mobile 3 cm right thyroid nodule that was firm, without cervical adenopathy. There was complete absence of the left breast and pectoralis muscles, with normal development on the right (Figure 2). The left hand was relatively small and exhibited syndactyly (Figure 3). The lower extremities were normal and symmetric (Figure 4).

Biochemical testing revealed normal thyroid function. Neck ultrasound revealed complete absence of the left thyroid lobe and the presence of a well-circumscribed hypoechoic 3 cm right thyroid nodule without calcifications, TIRADS score 3 (Figures 5 and 6). There was no abnormal cervical adenopathy. Chest X-ray revealed dextrocardia (Figure 7).

The patient was diagnosed with Poland syndrome, a 3 cm right thyroid nodule and agenesis of the left thyroid lobe. Her consent was obtained for a right thyroid lobectomy due to the cosmetic impact of the nodule, as well as the small risk that this represented a thyroid malignancy.

An uncomplicated right thyroid lobectomy was performed. In the operating room it was noted that the isthmus did not extend onto the left side of the neck, and did not require division. The patient did well and was discharged on the first postoperative day on thyroid hormone replacement. There was no evidence of hypoparathyroidism, and there were no postoperative complications. Final surgical pathology was consistent with a 3 cm benign follicular adenoma.

Discussion

Most descriptions of Poland syndrome characterise it as a rare congenital syndrome causing hypoplasia or agenesis of the pectoralis major. The first description was made in 1841 by Alfred Poland, then a student of anatomy.[2,5,6] The pathogenesis of Poland syndrome is not well understood. The most accepted theory postulates that a vascular accident during the sixth

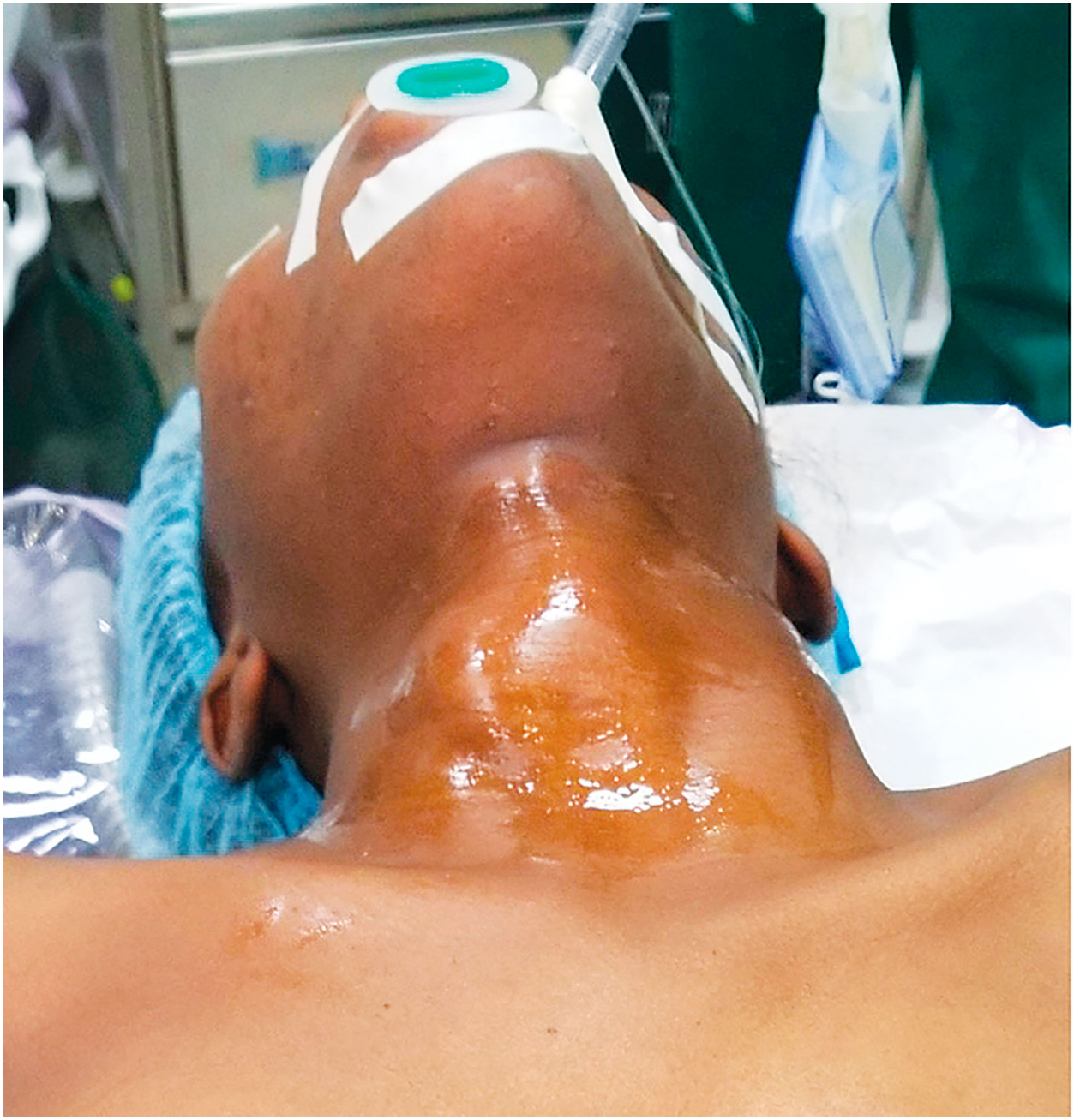

Figure 1: Chronic right neck swelling causing physical disfigurement

week of embryological development results in the disruption of blood flow in the subclavian and vertebral arteries and their branches on the affected side.[7] Most often the abnormality occurs on the right chest.

Frequently associated findings include ipsilateral mammary agenesis and syndactyly,[2] as in our patient. Thyroid hemiagenesis is an often asymptomatic congenital malformation, in which one thyroid lobe fails to form.[8] Most patients are euthyroid.[9] The first description was in 1895 by Marshall.[3] The pathogenesis of thyroid hemiagenesis remains obscure, most theories suggesting a genetic link, based on familial clusters.[10,11] The descent of the thyroid primordium from the primitive pharynx to its eventual location is closely associated with migration of the aortic sac and its branches. Therefore, many theorise that abnormal vascular development is the cause of thyroid hemiagenesis.[12,13] Agenesis of the left lobe is most frequent, occurring 68–80% of the time,[8] as in our patient.

Conditions reported to be associated with thyroid hemiagenesis have included parathyroid disorders, right aortic arch and Down syndrome.[14–17] Usually thyroid function is normal, but average TSH levels are higher than in patients with normal bilateral thyroid lobes.[18,19] Thus there is rarely a need for treatment, unless a patient develops hypothyroidism related to Hashimoto's disease. Elevated TSH levels are thought to lead to a compensatory increase in the size of the one lobe that is present, and some investigators have shown increased nodular disease, as was seen in our patient.[18]

Thyroid hemiagenesis is often discovered by ultrasound or scintigraphy during an evaluation of thyroid dysfunction or

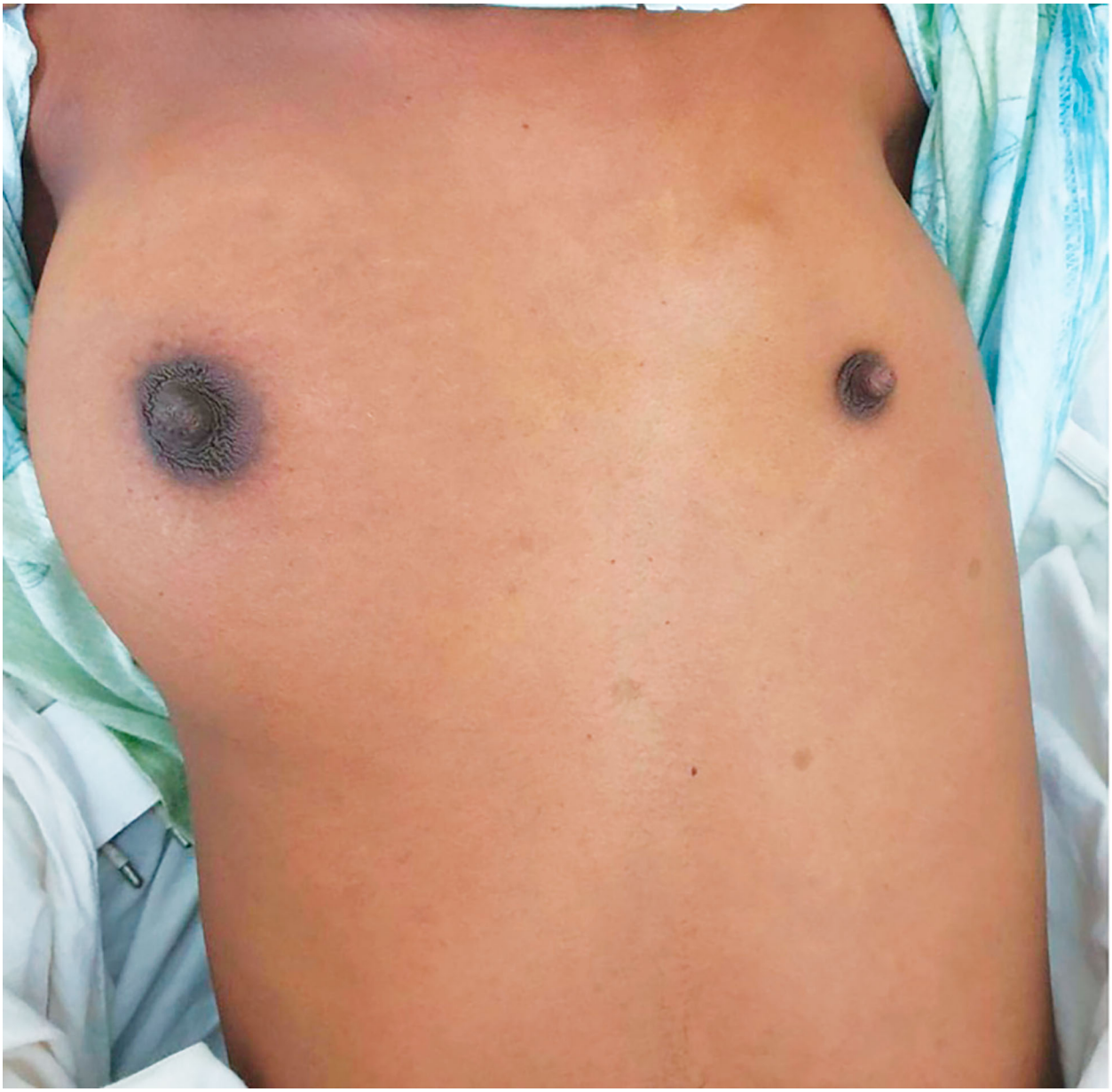

Figure 2: Absence of the left breast and pectoralis muscles, with normal development on the right

contralateral goitre, as in our patient.[4,8] Otherwise it may be discovered incidentally during cervical imaging for unrelated reasons.[20] Ultrasound is the examination of choice for evaluating the thyroid, as it is best able to characterise the nature of the thyroid parenchyma and nodular disease.[20,21]

In cases of hemiagenesis, it confirms the absence of one lobe and provides detailed information regarding the contralateral thyroid. It is also readily available and low in cost.[22,23] Both anomalies are quite rare, with Poland syndrome estimated at 1–3 cases per 100 000 people,[2,5,7] while thyroid hemiagenesis is thought to be present in 0.025–0.2% of the population.[19,24–26] Most studies more commonly report thyroid hemiagenesis in females, which was up to seven times more prevalent in the study by Ruchala et al.,[18] but there are reports of a slightly higher male predominance.[19]

Poland syndrome has been described in association with other congenital abnormalities including Sprengel deformity of the scapula, Klippel–Feil spinal abnormalities, as well as both Moebius and Adams–Oliver syndromes.[5] Association of left-

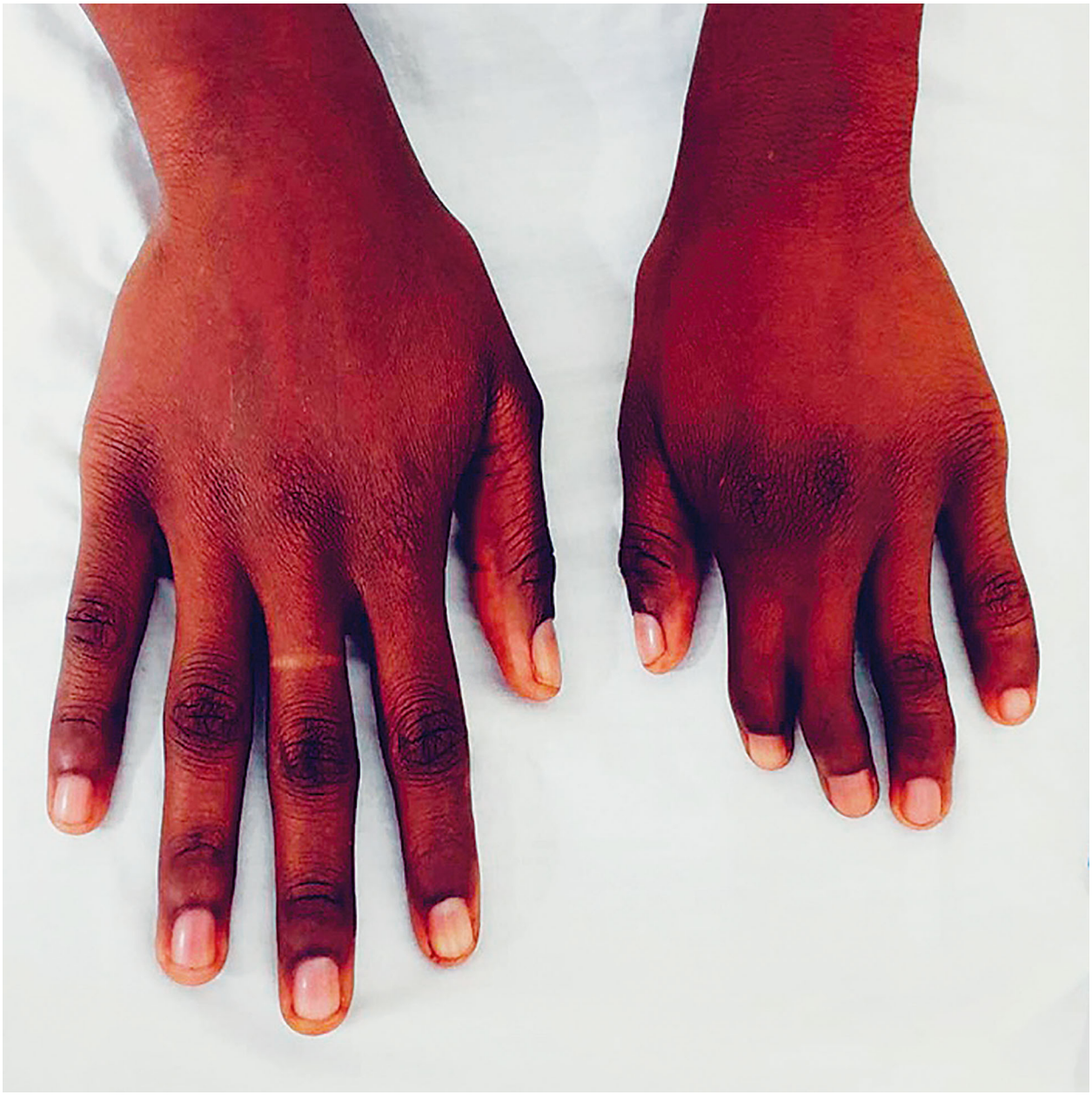

Figure 3: Small left hand exhibiting syndactyly

sided Poland syndrome and dextrocardia is not uncommon. The theory most cited as an explanation for this is that hemiagenesis of at least 2 ribs pushes the developing heart to the right.[27] Most often in cases of Poland syndrome in women, it is the lack of development of breast tissue that is most troubling for the patient, and ultimately prompts a consultation for cosmetic surgical repair.[1,5] However, in our patient, this was not the case, and her main aesthetic concern was the visible right-sided goitre.

Typically, thyroid lobectomy and isthmusectomy are performed for one of the following reasons: an FNA suggestive or diagnostic of thyroid carcinoma, a unilateral compressive nodule, a toxic adenoma, or a large nodule causing disfigurement. Usually, the approach to a nodule such as the one present in our patient would be to obtain a fine-needle aspiration (FNA) biopsy after assessment of thyroid function with a serum thyroid stimulating hormone (TSH) assay. However, biopsy was not performed in this case because the patient requested lobectomy for cosmetic reasons.

Conclusion

Poland syndrome and thyroid hemiagenesis are both rare congenital malformations. Their association has not been previously described. Since the theories behind the causation of each pathology remain unclear, this case may reveal an underlying association. Based on our findings, when evaluating patients with Poland syndrome and thyroid disease, an

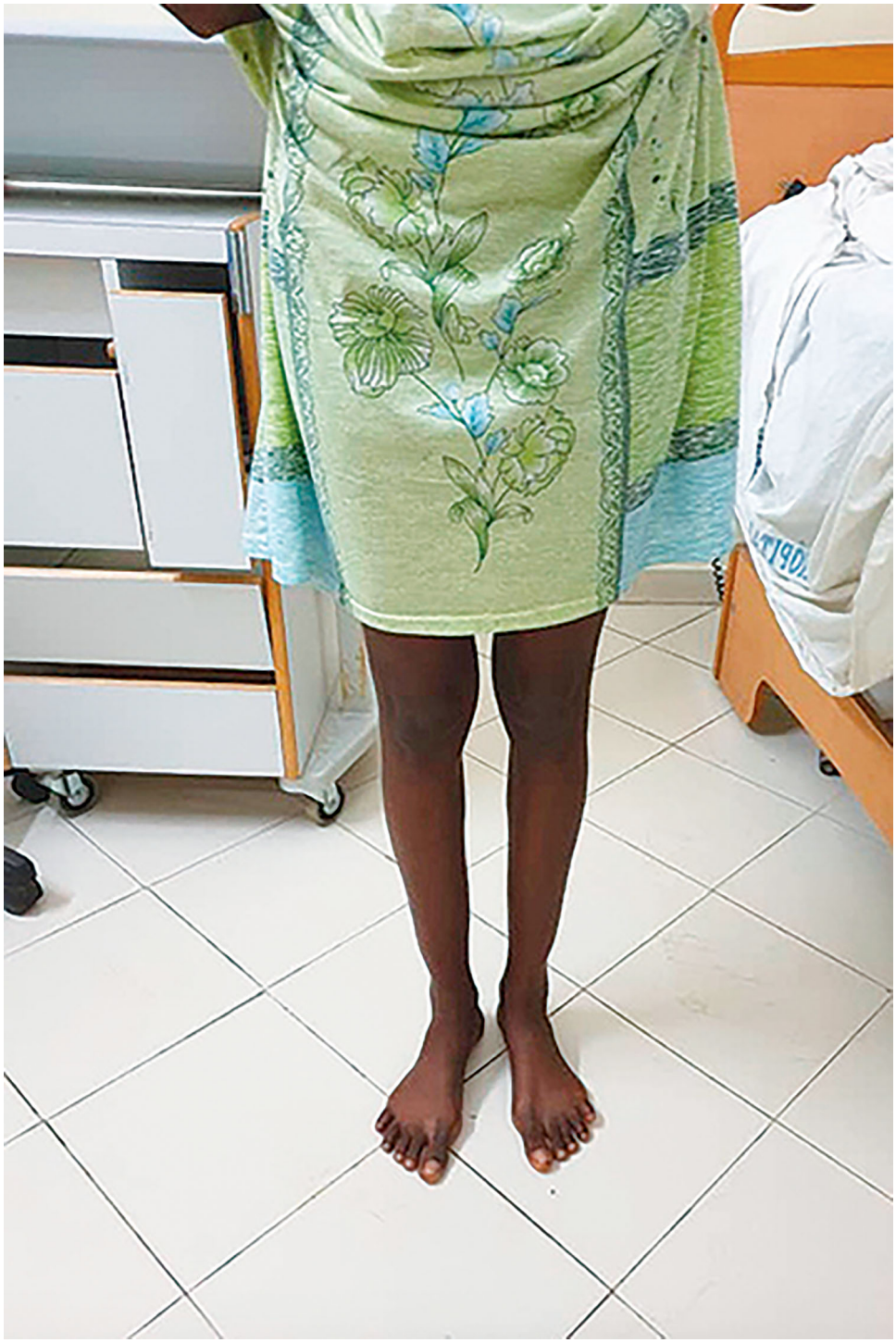

Figure 4: Normal, symmetric lower extremities

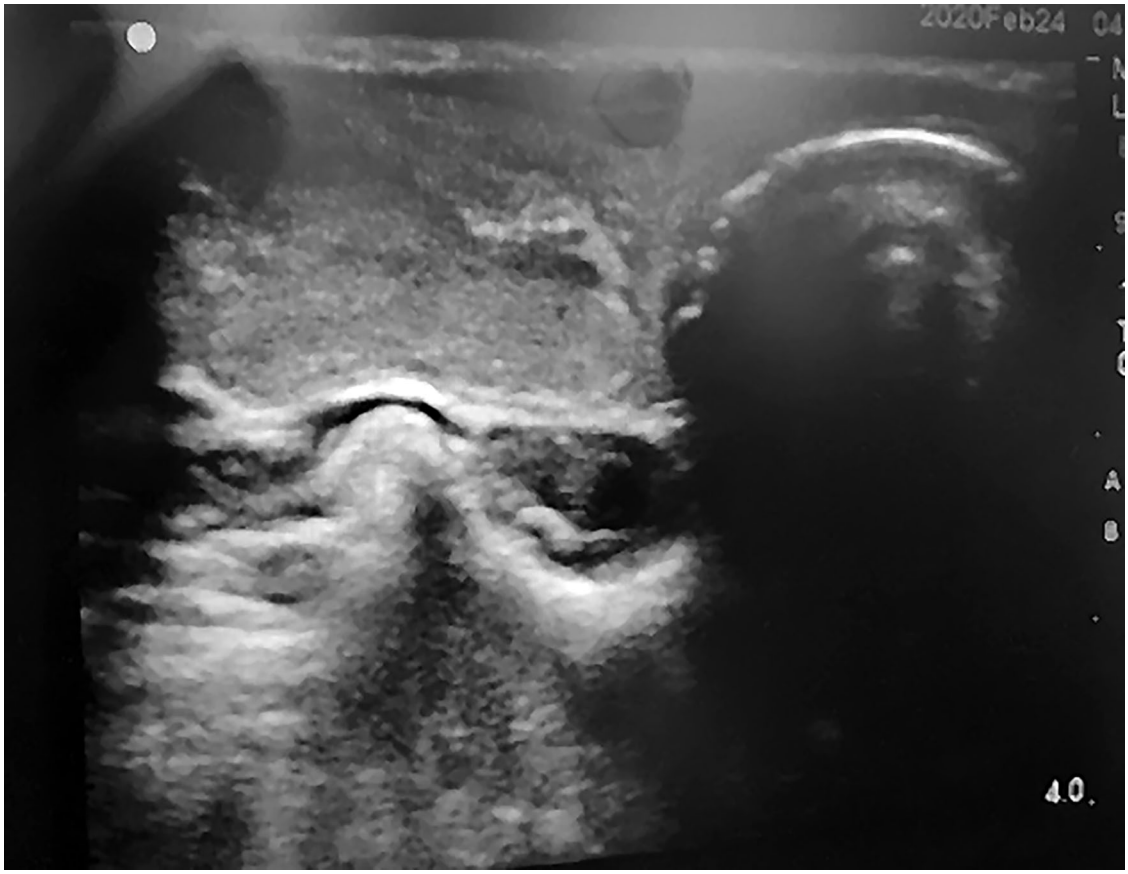

Figure 5: Neck ultrasound showing absence of the left thyroid lobe and the presence of a 3 cm hypoechoic right thyroid nodule without calcifications (front/back/side view)

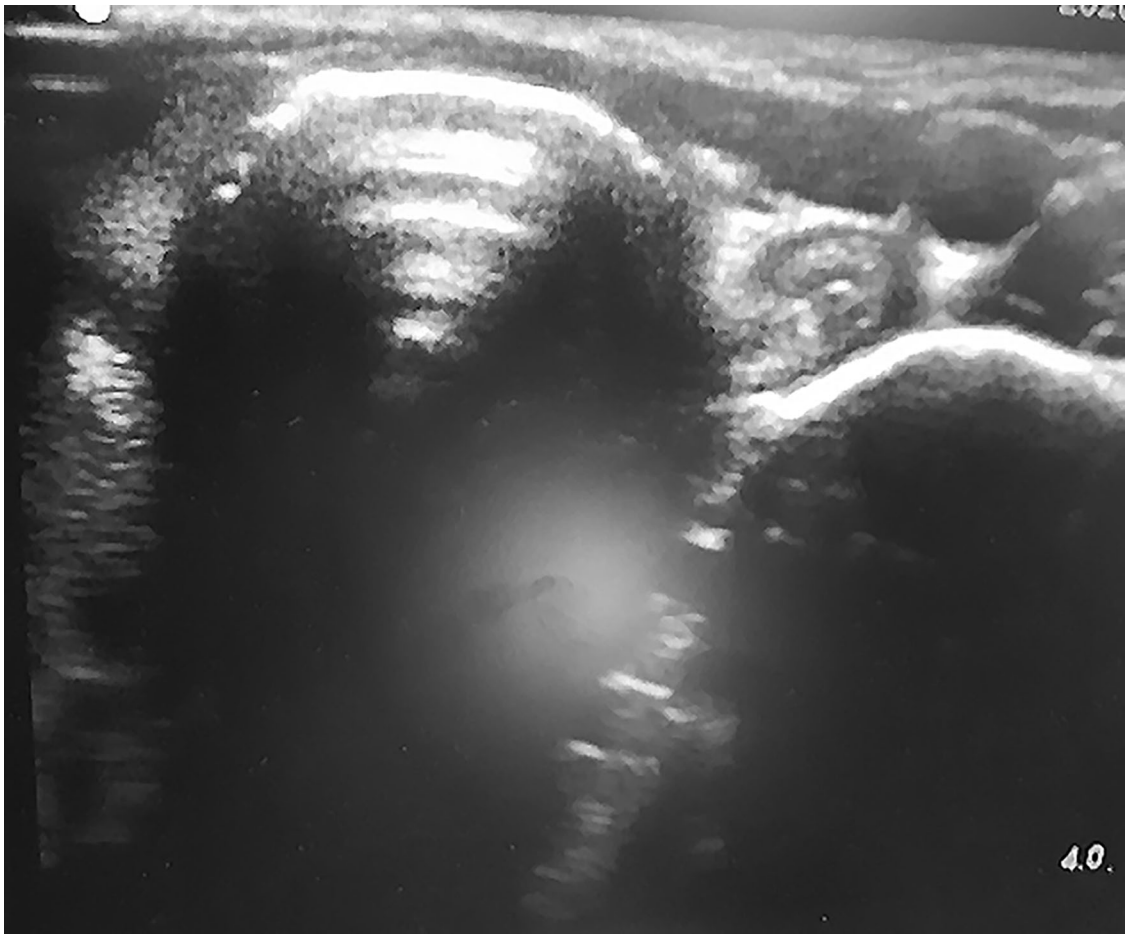

Figure 6: Neck ultrasound showing absence of the left thyroid lobe and the presence of a 3 cm hypoechoic right thyroid nodule without calcifications (front/back/side view)

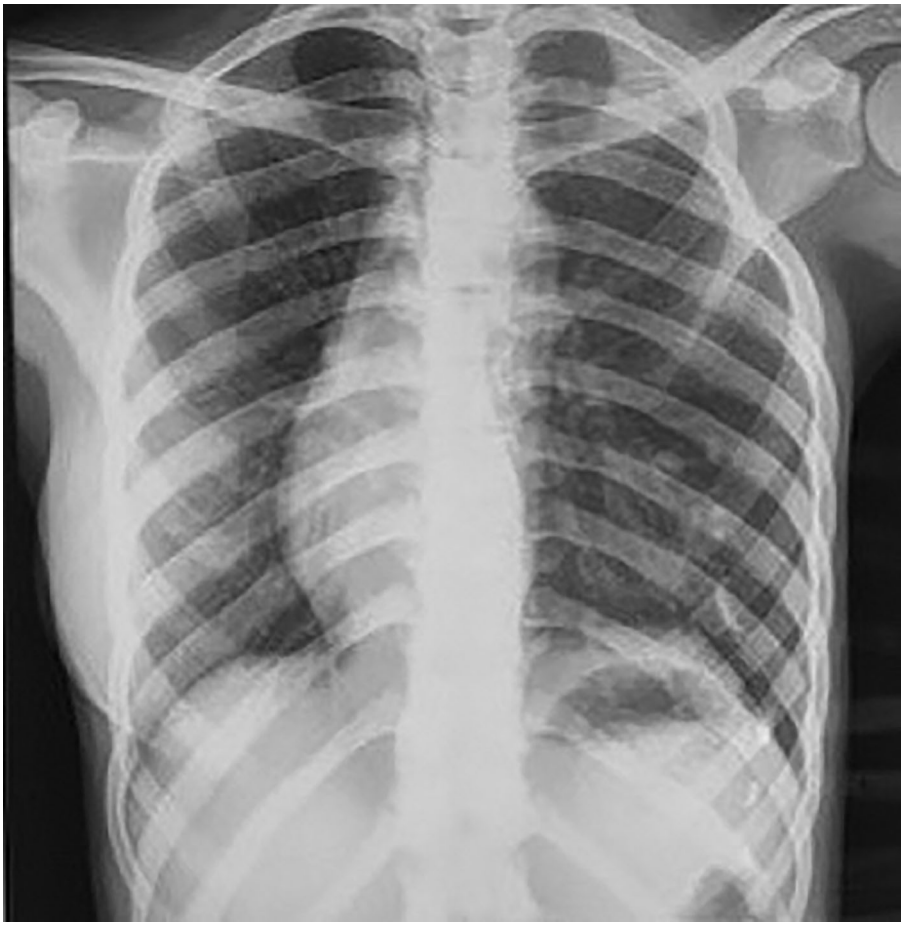

Figure 7: Chest X-ray showing dextrocardia

ultrasound of the thyroid should be obtained to assess for the possibility of hemiagenesis. If present, the patient must be informed prior to thyroid lobectomy of the mandatory need for lifelong thyroid hormone replacement. Of course, the ultrasound findings will also inform the medical team that such a patient needs to be started on thyroid hormone replacement postoperatively, unlike a typical lobectomy patient, who would be discharged without replacement and undergo TSH testing later.

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