

In this patient none of the recognized mutations known to cause Liddle's syndrome were found. However, the normal urinary steroid excretion profile and the response to amiloride but not to spironolactone is highly suggestive of unregulated activation of the renal tubular Na<sup>+</sup> transport, caused by an as-yet unidentified mutation in this family which is likely to have been transmitted with autosomal dominant inheritance.

This report demonstrates that patients with hypertension and hypokalaemia, without associated endocrine abnormalities, who respond to treatment with amiloride but not to spironolactone, are likely to have a diagnosis of Liddle's syndrome or its variant. Treatment with amiloride is safe, effective and likely to reduce arrhythmic events following acute myocardial ischaemia and infarction in these patients. **BJHM**

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- Cotton RGH, Scriver CR. Proof of "disease causing" mutation. *Hum Mutat.* 1998;12(1):1–3. [https://doi.org/10.1002/\(SICI\)1098-1004\(1998\)12:1<:AID-HUMU1>3.0.CO;2-M](https://doi.org/10.1002/(SICI)1098-1004(1998)12:1<:AID-HUMU1>3.0.CO;2-M)
- Kryukov GV, Pennacchio LA, Sunyaev SR. Most rare missense alleles are deleterious in humans: implications for complex disease and association studies. *Am J Hum Genet.* 2007 Apr;80(4):727–739. <https://doi.org/10.1086/513473>
- Liddle GW, Bledsoe T, Coppage WS. A familial renal disorder simulating primary aldosteronism but with negligible aldosterone secretion. *Trans Assoc Am Physicians.* 1963;76:199–213.
- Rossi E, Farnetti E, Debonneville A et al. Liddle's syndrome caused by a novel missense mutation (P617L) of the epithelial sodium channel beta subunit. *J Hypertens.* 2008 May;26(5):921–927. <https://doi.org/10.1097/HJH.0b013e3282f85dfe>
- Snyder PM, McDonald FJ, Stokes JB, Welsh MJ. Membrane topology of the amiloride-sensitive epithelial sodium channel. *J Biol Chem.* 1994 Sep 30;269(39):24379–24383.
- Wang W, Zhou W, Jiang L et al. Mutation analysis of SCNN1B in a family with Liddle's syndrome. *Endocrine.* 2006;29(3):385–390. <https://doi.org/10.1385/ENDO:29:3:385>

## LEARNING POINTS

- Early onset, resistant hypertension with hypokalaemia should prompt investigation for Liddle's syndrome and exclusion of other secondary causes of hypertension.
- Genetic analysis for Liddle's syndrome is indicated in patients with a family history of hypertension suggestive of autosomal dominant expression.
- Targeted treatment involves potassium-sparing diuretics, such as amiloride, which selectively block epithelial sodium channel activity. Conventional antihypertensive agents are ineffective.
- Response to amiloride without recurrence of hypertension and hypokalaemia supports a diagnosis of Liddle's syndrome.
- By normalizing blood pressure and correcting potassium levels, amiloride afforded a membrane-stabilizing effect, therefore reducing further arrhythmias.

## Images in Medicine

# A surprising chest radiograph

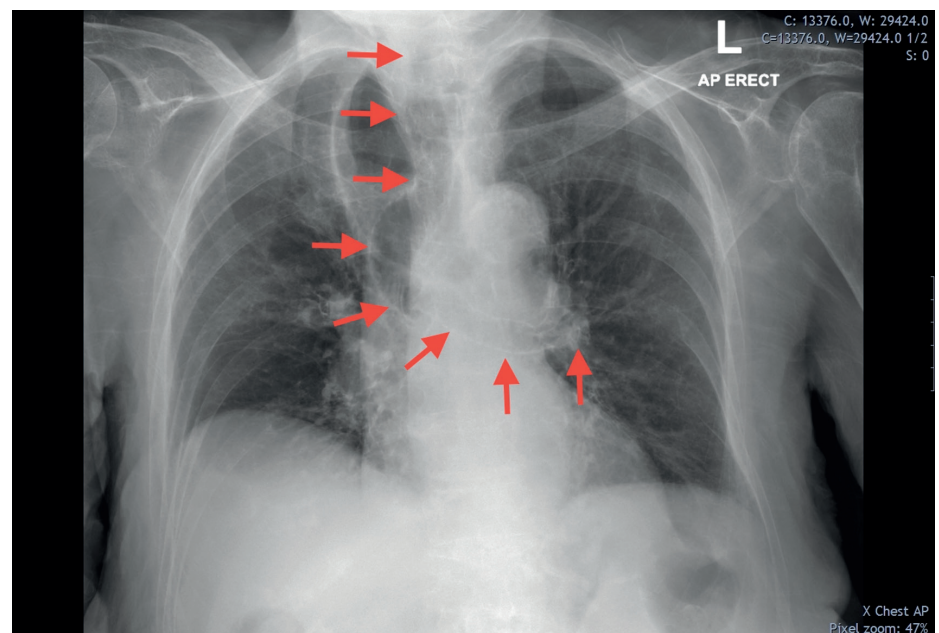
**A** 97-year-old woman's routine admission chest radiograph showed a peculiar undulating tubular structure overlying the cardiac shadow in the upper to mid thoracic zone. Clinical examination was unremarkable. The patient was noted to be producing copious amounts of white phlegm and was reported to have a reduced oral intake with no symptoms of dysphagia or regurgitation. A collateral history confirmed the diagnosis of achalasia.

Achalasia is a rare gastroenterological condition (Schlottmann and Patti, 2018),

and this case highlights an uncommon incidental finding of achalasia on a chest radiograph. **BJHM**

Schlottmann F, Patti MG. Esophageal achalasia: current diagnosis and treatment. *Expert Rev Gastroenterol Hepatol.* 2018 Jul;12(7):711–721. <https://doi.org/10.1080/17474124.2018.1481748>

**Figure 1. Chest radiograph showing achalasia. The abnormality is in the upper and mid zones; its course is mapped by the red arrows.**



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