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Aplasia Cutis Congenita on the Scalp

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INTRODUCTION

Aplasia cutis congenita is a rare condition characterized by localized absent skin at birth, often affecting the scalp. The exact pathogenesis is unclear, but it is likely related to disrupted prenatal skin development. Potential etiologies include genetic factors, trauma, intrauterine infections, teratogens, incomplete neural tube closure, and vascular compromise. We present a case of aplasia cutis congenita on the scalp potentially associated with maternal methimazole use.

A term male newborn was delivered by a 34-year-old female with a history of Graves' disease. The newborn had three well-demarcated, round lesions of absent skin on the scalp, measuring up to 1 centimeter in diameter. (Figure 1) The mother had been treated with methimazole 5 mg/day for mild Graves' hyperthyroidism. After 78 days of treatment, she tested positive for pregnancy. Methimazole was switched to propylthiouracil 50 mg/day at 9 weeks of gestation. She remained euthyroid, and propylthiouracil was discontinued at 23 weeks of gestation until delivery. The diagnosis of aplasia cutis congenita was made, potentially

Figure 1. Three well-demarcated, round scalp lesions characterized by full-thickness skin loss without involvement of the calvarium or signs of inflammation, each measuring up to 1 cm in diameter, consistent with aplasia cutis congenita on the scalp.

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related to methimazole exposure, known as methimazole embryopathy. Treatment involves simple wound care, leading to healing within a few months and often leaving an atrophic scar. Large lesions or those involving the skull may require surgical reconstruction.

In this patient, with deep scalp lesions located at the midline vertex, imaging studies are indicated to assess for underlying bone defects, vascular anomalies, or brain malformations such as meningomyelocele or porencephaly. Fortunately, the ultrasound revealed no abnormalities. Diagnosis is generally based on clinical assessment; a skin biopsy is not necessary in our case unless other lesions are suspected, and it should not be performed without prior imaging. The differential diagnosis includes conditions such as obstetric trauma from forceps, vacuum extraction, or fetal scalp monitor electrodes; infections like herpes simplex or varicella zoster; and epidermolysis bullosa. However, there is no evidence of these conditions in this case.

Current evidence does not establish a direct causal relationship between aplasia cutis congenita and methimazole use. However, antithyroid drugs should be avoided in the first trimester of pregnancy to prevent teratogenic effects, as both methimazole and propylthiouracil cross the placenta.[1] The 2017 American Thyroid Association Guidelines recommend using propylthiouracil in the first trimester and switching to methimazole in the second trimester. This approach helps mitigate the risk of hepatotoxicity associated with propylthiouracil while effectively managing hyperthyroidism, and reduces the teratogenic risks of methimazole, which can cause more severe birth defects than propylthiouracil, as major organs and systems are formed by the end of the first trimester.[2,3] For patients with mild hyperthyroidism, propylthiouracil may be considered before conception, and antithyroid drugs can be discontinued upon confirming pregnancy if thyroid function remains normal.[2,3]

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Ethical Consideration
The patient consent form was obtained before manuscript submission.

Statement of Authorship
All authors certified fulfillment of ICMJE authorship criteria.

CRediT Author Statement
SW: Conceptualization, Investigation, Resources, Data Curation, Writing – original draft preparation, Writing – review and editing, Visualization; **CS:** Conceptualization, Investigation, Resources, Data Curation, Writing – review and editing, Visualization, Supervision, Project Administration.

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