Aplasia Cutis Congenita Following Maternal Carbimazole Use in Pregnancy: First Case from Oman

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Dear Editor,

A 5-day-old Omani male neonate presented to our dermatology clinic with localized scalp lesions present since birth. Antenatal history was significant for maternal hyperthyroidism, specifically Graves' disease. The mother had been prescribed Carbimazole (CMZ) as an antithyroid medication three years previously. Thyroid function tests (TFTs), including thyroid-stimulating hormone (TSH), remained within normal range throughout her pregnancy. The infant was delivered at term via uncomplicated vaginal birth to non-consanguineous parents. Notably, the patient's 3-year-old sister had been diagnosed at birth with aplasia cutis congenita (ACC) during the same period in which their mother began CMZ treatment, while his two eldest siblings were unaffected. The patient was evaluated a few days after delivery by a multi-disciplinary team comprising dermatologists, a pediatrician, and cardiologists. Upon examination, he appeared well, no abnormal movements or dysmorphic features. Local examination of the scalp revealed two areas of skin and subcutaneous tissue loss at the vertex, measuring 0.5 x 0.5 cm and 4 x 4 cm, respectively. Tissue loss did not extend deep into the fascia and no discharge was noted [Figure 1]. Systemic examination was unremarkable.

Figure 1: (**A & B**) Two ACC lesions over the scalp.

Laboratory tests were done included a cord TSH test, which was normal. On the second day of life, TFTs showed TSH at 46.9 uIU/mL (reference range: 0–20 uIU/mL), and free thyroxine (T4) at 30.1 pmol/L (reference range: 11–32 pmol/L). However, by the fourth day of life, TSH and free T4 levels had dropped to 5.2 uIU/mL and 33.7 pmol/L, respectively. Brain ultrasonography was unremarkable.

The final diagnosis was of ACC likely secondary to maternal use of CMZ during pregnancy. The patient underwent conservative management, consisting of a topical antibiotic (mupirocin) to prevent secondary bacterial infections and appropriate wound care to allow the skin defects to heal by themselves. By a three-month follow-up, the scalp defects had completely healed, leaving hairless atrophic scars [Figure 2]. No additional complications were recorded.

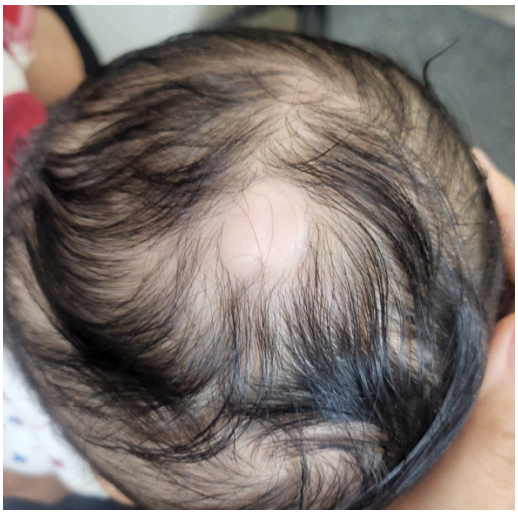

Figure 2: ACC healed with hairless atrophic scars.

Aplasia cutis congenita (ACC) is a rare congenital skin defect characterized by the absence of skin and, in some cases, underlying subcutaneous tissue, bone, or dura.[1] The pathogenesis of ACC is linked to various factors, such as genetic predisposition, prenatal trauma, vascular impairment, infection, and teratogenic exposure to various medications, such as valproic acid and antithyroid medications.[2] Diagnosis of ACC is based on clinical presentation, as histology features provide limited diagnostic value.[1, 2]

Over the past four decades, multiple case reports have suggested a link between maternal exposure to CMZ or MMI and ACC. A case series and literature review by Sachs et al. documented 61 cases of ACC linked primarily to MMI/CMZ exposure.[3] This entity, known as MMI/CMZ embryopathy, can present as isolated ACC or as part of a broader embryopathy with anomalies like choanal atresia, omphalocele, and esophageal atresia.[4] The absolute risk of MMI/CMZ embryopathy following first-trimester exposure to these drugs is estimated to be 1.6–3% [5]. A meta-analysis of two large retrospective studies and six cohort studies reported that prenatal MMI/CMZ exposure increased the risk of congenital abnormalities, including ACC, by 64%.[5] Some researchers have proposed that underlying maternal thyrotoxicosis is the main culprit of such defects; however, other cases, including ours, have documented ACC in neonates of euthyroid mothers.[5] Professional guidelines recommend preconception counseling for women requiring antithyroid treatment. In addition, experts advise switching to propylthiouracil (PTU) in the first trimester—when organogenesis is most critical—and resuming MMI/CMZ as of mid-pregnancy after initial fetal organ development is complete.[4]

There is currently no consensus regarding the optimal threshold and specific treatment algorithm for management. Strategies range from conservative care to surgical intervention based on factors such as lesion location, size, underlying causes, and associated anomalies.[6] Our patient underwent conservative therapy because the lesions were relatively small (diameter: 1.5–2.0 cm) with no signs of infection or bone involvement. Rarely, complications, such as local infection, hemorrhage, meningitis, or sagittal sinus thrombosis, may occur that can worsen prognosis.[7] In our case, no complications arose, and healing was satisfactory.

Our case supports existing evidence linking exposure to MMI/CMZ during pregnancy with congenital malformations such as ACC, although a definitive causal relationship has yet to be established. Given the recent recognition of MMI/CMZ embryopathy, there is a need for larger epidemiological studies to quantify the risk of congenital deformities among pregnant women taking antithyroid medications.

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