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# Sacral Agenesis Through a Preventive Medicine Lens: Maternal Risk, Early Detection, and Lifelong Health Implications

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ABSTRACT

Sacral agenesis, a rare congenital malformation within the caudal regression spectrum, is characterized by partial or complete absence of the sacrum and is frequently accompanied by multisystem involvement affecting neurologic, orthopedic, urologic, and gastrointestinal function. Despite its low incidence, sacral agenesis is associated with substantial lifelong morbidity and healthcare utilization, underscoring its relevance beyond rarity alone. Robust epidemiologic evidence demonstrates a strong association between maternal pregestational diabetes and sacral agenesis, identifying a modifiable risk factor that places this condition squarely within the domain of preventive medicine. Advances in prenatal imaging allow for antenatal detection, yet disparities in access and delayed diagnosis limit opportunities for anticipatory care and complication prevention. This editorial argues for reframing sacral agenesis as a preventable and risk-modifiable condition through improved preconception metabolic health, equitable prenatal screening, and coordinated lifelong care. Viewing sacral agenesis through a preventive medicine lens highlights actionable strategies to reduce avoidable morbidity and advance health equity for affected individuals and families.



Sacral agenesis, also known as caudal regression syndrome, is a rare congenital condition characterized by partial or complete absence of the sacrum and often associated with additional anomalies of the spinal cord and lower extremities [1]. This condition's multisystem impact spans orthopedic, genitourinary, gastrointestinal, and neurologic domains, contributing to lifelong morbidity [2][3]. Although sacral agenesis affects approximately 2.6 per 100,000 live births, population-based surveillance shows significant associations with modifiable maternal exposures [4]. Current clinical discourse tends to emphasize surgical and rehabilitative care, overlooking opportunities for upstream preventive interventions. Repositioning sacral agenesis within a preventive medicine framework foregrounds maternal health, early detection, and health system responsiveness as levers for reducing avoidable morbidity.

One of the strongest risk factors for sacral agenesis is maternal pre-gestational diabetes—including both type 1 and type 2 diabetes prior to conception—as shown by adjusted odds ratios which far exceed those associated with any other exposures [4]. Diabetic embryopathy describes, in broad terms, the congenital anomalies produced by maternal hyperglycemia, including caudal regression spectrum defects [2][5]. Risk is dramatically increased for women with poor glucose control during the periconceptional period, making optimization of a woman's metabolic status before conception a modifiable risk factor. Smoking during the periconceptional period is also a risk factor for women without diabetes in multivariable analyses and is another point of intervention for counseling and education [4]. Because there is a multifactorial aetiology to sacral agenesis, including sporadic genetic and environmental components and various vascular anomalies, it is unlikely that the prevention of sacral agenesis can occur solely through the management of diabetes. Thus, primary prevention strategies should include broad-based health promotion activities aimed at addressing each of the risk factors identified in the literature.

Antenatal detection of sacral agenesis through routine obstetric ultrasound and confirmatory fetal magnetic resonance imaging is feasible, enabling earlier prognostication and clinical planning [6]. Prenatal diagnosis provides a critical window for interdisciplinary counseling, risk stratification, and preparation for neonatal needs, all of which align with preventive care paradigms aimed at reducing avoidable complications. However,

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disparities in access to high-quality imaging and specialist evaluation compromise equitable early identification, especially in underserved settings [3]. A preventive medicine approach would prioritize standardized screening protocols and streamlined pathways to expert referral to ensure that detection leads to actionable care planning rather than delayed or fragmented follow-up. Integrating sacral agenesis into broader prenatal risk assessment frameworks underscores the need for surveillance systems that capture rare congenital anomalies for quality improvement and prevention research.

When primary prevention is not achievable, secondary and tertiary prevention—aimed at mitigating complications and preserving function—becomes essential for individuals with sacral agenesis. Because of its frequent association with neurogenic bladder, bowel dysfunction, lower limb anomalies, and other sequelae, early multidisciplinary involvement can reduce preventable morbidity [7]. Longitudinal care coordination, including urology, orthopedics, rehabilitative medicine, and social support services, exemplifies tertiary prevention that prevents secondary disability. Fragmented care delivery and loss to follow-up undermine these preventive goals, particularly during transition from pediatric to adult health systems. A preventive medicine strategy prioritizes continuity of care, structured long-term surveillance, and integration of patient-reported outcomes to anticipate and avert complications before they escalate.

Sacral agenesis, while rare, highlights systemic gaps at the intersection of maternal health, prenatal care, and lifelong preventive care that are applicable to many congenital disorders. Integrating congenital anomaly prevention into chronic disease management policies for women of reproductive age—particularly diabetes care—would align maternal health systems with preventive objectives. Public health programs should promote preconception counseling, glycemic optimization, cessation of harmful exposures, and equitable access to prenatal imaging as core elements of a congenital anomaly prevention suite. Health systems and policymakers must also invest in registries and surveillance infrastructure that track rare conditions like sacral agenesis to inform quality improvement and research. By reframing sacral agenesis through the lens of preventive medicine, clinicians and public health leaders can reduce avoidable morbidity and enhance health equity for affected individuals and families.

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