Polydactylism in Cattle: Genetic Basis, Clinical Significance and Implications for Breeding

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ABSTRACT

Polydactylism is a rare congenital anomaly characterized by the presence of one or more extra digits in cattle. The condition has been reported in several breeds, including Holstein, Simmental, Dexter, Hereford, and Angus, and is generally considered hereditary. Although its exact mode of inheritance remains unclear, evidence suggests involvement of incomplete penetrance, variable expressivity, and polygenic influences. Genes associated with embryonic limb development, particularly those within the Sonic Hedgehog (SHH) signaling pathway, including SHH, LMBR1, GLI3, and HOXD13, are considered important candidates. Clinical manifestations range from minor cosmetic defects to severe locomotor impairment. Polydactylism is of economic and breeding significance because affected animals are typically excluded from breeding programs to prevent transmission of the defect. Despite reports in several cattle breeds worldwide, documented cases in indigenous Indian cattle breeds remain scarce. Advances in genomics may help elucidate the molecular basis of this anomaly and facilitate its management through informed breeding strategies.

INTRODUCTION

Polydactylism is a congenital developmental anomaly characterized by the presence of one or more extra digits (toes) beyond the normal two functional digits in cattle. It is an uncommon hereditary

defect that can affect one or more limbs and may occur unilaterally or bilaterally.

Types of Polydactylism

1. Preaxial polydactylism – Extra digit occurs on the medial (inner) side of the limb.
2. Postaxial polydactylism – Extra digit occurs on the lateral (outer) side of the limb.
3. Central polydactylism – Additional digit develops between the normal digits (rare).

Genetic Basis of Polydactylism

1. Hereditary Inheritance

Studies in Simmental, Holstein, and Dexter cattle suggest that polydactylism is inherited, but the mode of inheritance appears complex:

- Autosomal dominant with incomplete penetrance has been proposed in some families.
- Polygenic inheritance was suggested in Simmental cattle because the defect did not follow a simple Mendelian pattern.
- Variable expressivity is common; affected animals may show different numbers and locations of extra digits.

2. Candidate Genes

Several genes involved in embryonic limb development are considered candidates:

Gene	Function	Relevance to Polydactyly
SHH (Sonic Hedgehog)	Controls anterior-posterior limb patterning	Major gene implicated in polydactyly across many species
LMBR1	Contains the ZRS (Zone of Polarizing Activity Regulatory Sequence)	Mutations frequently cause preaxial polydactyly
	enhancer controlling SHH expression	
GLI3	Mediates SHH signaling	Mutations associated with extra digits
HOXD13	Limb morphogenesis and digit formation	Developmental abnormalities including polydactyly
FGF family genes	Limb bud growth and differentiation	Abnormal signaling can affect digit number

Mechanism

During embryonic development, the Zone of Polarizing Activity (ZPA) regulates digit formation through SHH signaling. Abnormal activation of SHH on the anterior side of the limb bud can induce formation of additional digits.

A simplified mechanism is:

Mutation in ZRS/LMBR1 → Ectopic SHH expression → Altered limb patterning → Supernumerary digit formation

Clinical Features

Polydactylism is characterized by the presence of one or more supernumerary digits in addition to the normal digits. The extra digits may be fully developed and functional or rudimentary and non-functional. Clinical severity varies considerably, ranging from minor cosmetic abnormalities to significant locomotor impairment. While some affected animals remain asymptomatic, others may exhibit lameness, abnormal gait, claw deformities, and reduced mobility, depending on the location and degree of digit development.

Diagnosis

Diagnosis of polydactylism is primarily based on physical examination, which readily

identifies the presence of supernumerary digits. Radiographic evaluation is useful for assessing the number, structure, and anatomical attachment of the additional bones and digits. In suspected hereditary cases, pedigree analysis can aid in determining inheritance patterns and identifying affected bloodlines.

Economic and Breeding Significance

Polydactylism is generally regarded as an undesirable hereditary defect in cattle breeding programs. Affected animals are often excluded from breeding to minimize the risk of transmitting the condition to subsequent generations. In severe cases, the anomaly may impair locomotion, compromise animal welfare, and adversely affect productivity, thereby resulting in economic losses to livestock producers.

Treatment

Mild cases may require no intervention. Surgical removal of non-functional extra digits can be performed when the defect causes lameness or management problems.

Example

Polydactylism in cattle has been documented in several breeds, including Holstein, Holstein-Friesian, Simmental, Dexter, Hereford, and Angus cattle. The condition is generally considered hereditary, with studies in Simmental cattle suggesting a complex polygenic mode of inheritance, while pedigree analyses in Holstein and Dexter cattle support genetic transmission of the defect.

Breed	Reported Case	Major Findings
Holstein	Bähr *et al.* (2003)	Female German Holstein twin calf with polydactyly in both forelimbs and one hind limb; additional medial digits present.
Simmental	Johnson *et al.* (1981)	Hereditary polydactyly investigated through breeding trials; evidence suggested a polygenic mode of inheritance.
Simmental	Altenbrunner-Martinek *et al.* (2007)	Seven-week-old Austrian Simmental calf with bilateral preaxial polydactyly affecting both forelimbs.
Dexter	Browning *et al.* (2020)	First reported case in Dexter cattle: a cow and her daughter both exhibited polydactyly, supporting hereditary transmission.

Although polydactylism has been reported sporadically in several cattle breeds worldwide, including Holstein, Simmental, Hereford, Angus, and Dexter cattle, published reports in recognized Indian indigenous cattle breeds remain extremely rare. No well-documented cases have been identified in major Indian breeds such as Gir, Sahiwal, Tharparkar, Ongole, Kankrej, Hariana, Hallikar, or Red Sindhi in the readily accessible scientific literature.

The absence of published reports of polydactylism in Indian cattle breeds does not necessarily mean that the condition does not occur. The apparent absence of polydactylism in Indian indigenous cattle breeds is more likely due to a combination of underreporting, limited pedigree documentation, lower research intensity, and possible differences in genetic architecture, rather than definitive evidence that the defect does not occur in these breeds. Systematic genomic surveillance of indigenous cattle populations may reveal previously undocumented congenital anomalies, including rare limb malformations such as polydactylism.

CONCLUSION

Polydactylism is a rare congenital limb anomaly in cattle characterized by the presence of one or more supernumerary digits. Although reported only sporadically in breeds such as Holstein, Simmental, Dexter, Hereford, and Angus cattle, available evidence supports a hereditary basis for the condition. The molecular mechanisms are believed to involve disturbances in embryonic limb patterning, particularly alterations in the SHH signaling pathway and its regulatory elements such as the ZRS enhancer within LMBR1. However, the precise causative mutations responsible for bovine polydactylism remain largely unidentified. Advances in genomic technologies, including whole-genome sequencing and functional genomics, are expected to improve our understanding of the genetic architecture underlying this defect. Enhanced surveillance and genetic characterization of congenital anomalies in both exotic and indigenous cattle populations will contribute to more effective breeding strategies and improved animal welfare.

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