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Suspected 38,XY/38,XY Chimerism in a Male Tortoiseshell Cat with Piebaldism and Polydactyly: A Case Report

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Abstract: *This report describes a rare case of a male tortoiseshell cat affected by polydactyly and suspected 38,XY/XY chimerism. The cat exhibited tortoiseshell coloration with tabby markings, piebaldism affecting the pectoral region and distal tail, and bilateral symmetrical polydactyly involving all four limbs, with six digits on each thoracic limb and five on each pelvic limb. Cytogenetic investigations included conventional karyotyping and fluorescence in situ hybridization (FISH), complemented by polymerase chain reaction (PCR) targeting the SRY and AR genes. Conventional cytogenetic analysis revealed a normal male karyotype (38,XY), with no evidence of sex chromosome aneuploidy. PCR analysis confirmed the presence of both SRY and AR sequences. The association between the tortoiseshell phenotype and the normal male karyotype suggests possible 38,XY/XY chimerism, although genetically distinct cell populations were not demonstrated. The concurrent presence of polydactyly represents an additional congenital morphological anomaly. This case emphasizes the relevance of cytogenetic and molecular investigations in the evaluation of atypical sex-linked pigmentation phenotypes in domestic cats.*

Introduction

Coat color and white spotting are linked to genetic determinants like the Dominant White locus (W) [1]. Congenital variations such as polydactyly and syndactyly rarely present with complex traits. Tortoiseshell pigmentation involves X-linked Orange variation and X-inactivation [2]. Male tortoiseshell cats may show aneuploidy or chimerism [3], and a normal 38,XY blood karyotype alone cannot prove two distinct lineages [4]. Piebaldism is a pigmentary disorder characterized by depigmented patches of skin and hair resulting from defective migration of neural crest-derived melanocytes, and has been associated with mutations in genes including *EDN3, EDNRB, KIT, MITF, PAX3, SNAI2* or *SOX10*.

Materials and methods

Supplied photographs and the FCA115 laboratory report were reviewed. Methods included Giemsa cytogenetics, feline X/Y FISH probes, and PCR for *SRY* and *AR* with male, female and water controls. Multi-tissue STR/SNP genotyping, limb radiographs and reproductive data were unavailable.



Fig. 1. Stark: (A) tortoiseshell/tabby coat; (B) white distal tail; (C) forepaw appearance

The cat showed tortoiseshell coloration with tabby markings and pectoral/distal-tail white spotting (Fig. 1). The case description records six digits per thoracic limb and five per pelvic limb (22 total); radiographs were unavailable. Blood analysis reported normal 38,XY in all examined cells: 75 assessed by FISH and 35 by conventional cytogenetics. Seven cells were karyotyped; some were included as representative images (Fig 2-3).

Results and discussion

Karyotype and FISH support 38,XY in examined cells (Figs. 2–3). PCR detected SRY and AR (Fig. 4). These tests identify targeted chromosomes/sequences, not two distinct genomes. In a published 38,XY/38,XY chimera, confirmation required multi-tissue microsatellite analysis [4].

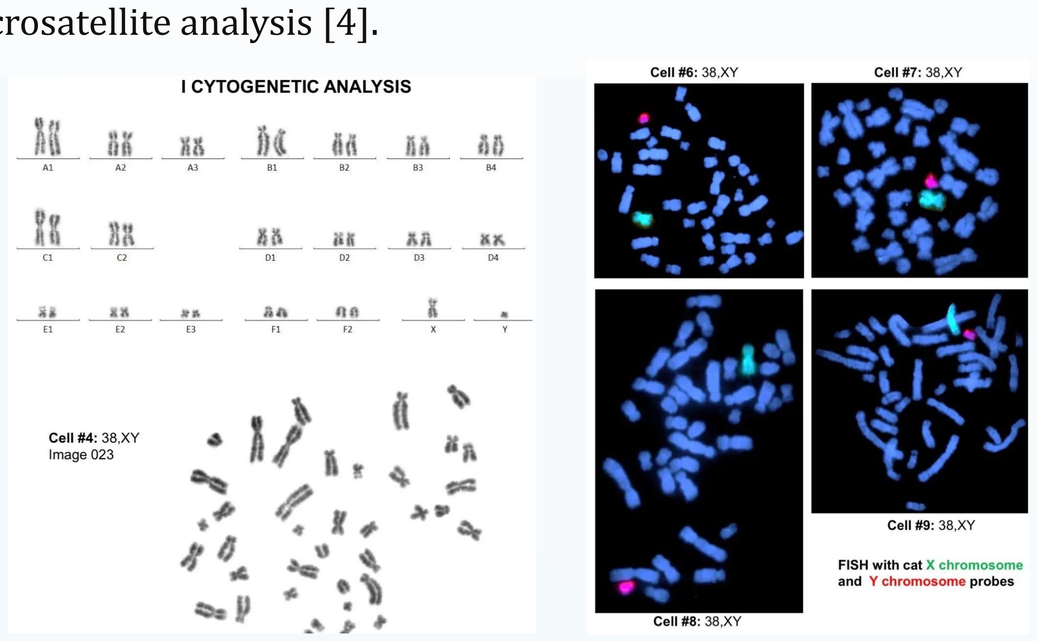


Fig. 2. Cell #4 (Image 023): 38,XY karyotype.

Fig. 3. FISH, Cells #6–#9: X green / Y magenta.

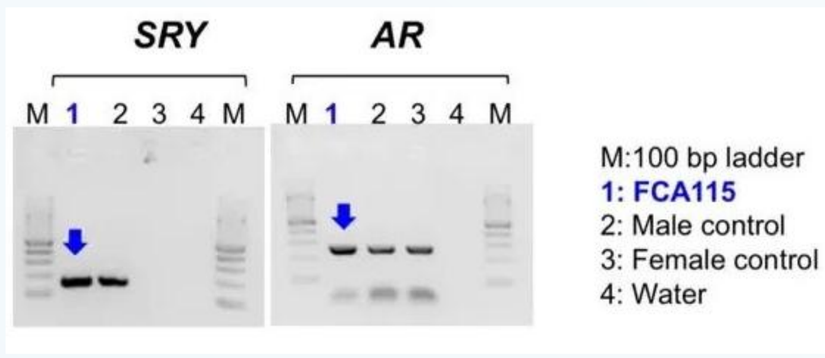


Fig. 4. SRY/AR PCR: M, 100-bp ladder; lane 1, FCA115; 2, male; 3, female; 4, water.

White spotting, tabby patterning, dilution and polydactyly involve distinct genetic pathways. ZRS lies within *LMBR1* and regulates *SHH* at a distance. None of these loci was genotyped in this case.

Conclusions

The findings support normal 38,XY in sampled blood cells and suspected—not confirmed—38,XY/38,XY chimerism. Multi-tissue lineage genotyping is required for confirmation. Fertility and specific pigmentation or polydactyly variants cannot be inferred from the available material.

References

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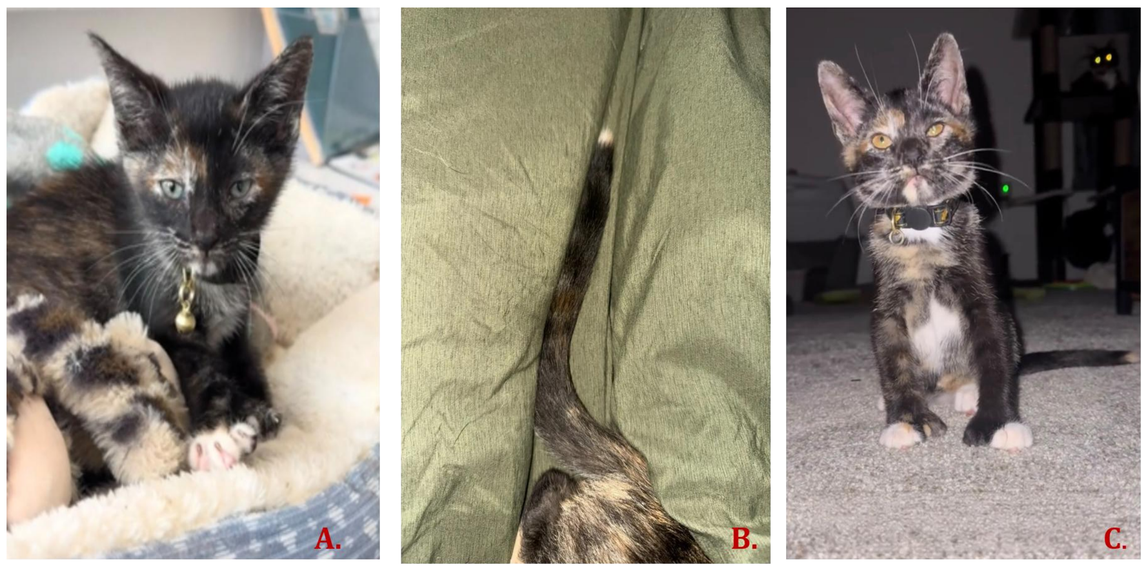


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