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AI-DRIVEN INSIGHTS INTO THE ROLE OF THE ACTN3 GENE IN MUSCLE STRENGTH AND SPRINT PERFORMANCE

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ABSTRACT

The ACTN3 gene, encoding the alpha-actinin-3 protein, plays a pivotal role in determining muscle performance, particularly in strength and sprint activities. The R577X polymorphism in the ACTN3 gene creates genetic variations that correlate with fast-twitch muscle fiber composition, influencing power, speed, and endurance. This study investigates the relationship between ACTN3 genotypes and athletic performance using Artificial Intelligence (AI). AI tools, including machine learning algorithms, predictive analytics, and data visualization, were employed to analyze a dataset of athlete profiles. Results reveal significant correlations between R allele carriers and enhanced sprint performance, while X allele homozygotes exhibited traits favoring endurance. AI-enabled models provided accurate performance predictions and uncovered complex gene-environment interactions. These findings highlight the transformative potential of AI in advancing sports genetics, paving the way for personalized training and talent identification strategies.

Keywords: ACTN3 Gene, Alpha-Actinin-3, R577X Polymorphism, Fast-Twitch Muscle Fibers, Sprint Performance, Muscle Strength, Artificial Intelligence, Machine Learning, Sports Genetics, Predictive Modeling, Talent Identification, Personalized Training, Gene-Environment Interaction, Athlete Performance Optimization

1. INTRODUCTION

The **ACTN3 gene** has emerged as one of the most extensively studied genetic markers in sports performance due to its critical role in determining muscle function. It encodes the **alpha-actinin-3 protein**, which is predominantly expressed in **fast-twitch muscle fibers**. These fibers are responsible for **high-speed and explosive power activities**, making the ACTN3 gene a significant factor in athletic capabilities.

The **R577X polymorphism** in the ACTN3 gene leads to two distinct alleles:

- **R allele**: Produces functional alpha-actinin-3 protein, associated with superior muscle strength and sprint performance.
- **X allele**: Results in the absence of alpha-actinin-3 protein, favoring traits beneficial for endurance-based activities.

This genetic variation influences muscle composition and functionality, directly affecting athletic performance across various sports disciplines.

Recent advancements in **Artificial Intelligence (AI)** have revolutionized the field of sports genetics, enabling researchers to analyze complex genetic datasets with **unprecedented accuracy and efficiency**. AI provides tools for uncovering intricate relationships between genetic variations, environmental influences, and performance outcomes. By leveraging machine learning models and predictive analytics, AI has the potential to offer deeper insights into the role of ACTN3 genotypes in sports performance, optimize training regimens, and enhance talent identification processes.

This study aims to utilize AI to explore the **influence of ACTN3 genotypes** on muscle strength and sprint performance, providing actionable insights into the genetic basis of athletic potential and its practical applications in sports science.

2. PURPOSE OF THE STUDY

The primary objectives of this study are:

- To investigate the relationship between ACTN3 gene variants and athletic performance metrics (strength and sprinting ability).
- To employ AI tools for enhanced analysis of genetic and performance data.
- To identify potential applications of AI-driven insights in personalized training and talent identification.

3. METHODOLOGY

This study involved 150 athletes (aged 18–35), categorized into sprint and endurance groups based on their performance metrics.

1) **Participants**
   - A cohort of 150 athletes (aged 18–35 years) specializing in sprint and endurance sports were selected.
   - Participants were categorized based on their performance profiles and sporting discipline.
2) **Genotyping**
   - DNA samples were collected via buccal swabs and analyzed for the R577X polymorphism using PCR techniques.
3) **Performance Metrics**
   - **Muscle Strength**: Assessed through one-repetition maximum (1RM) tests for key muscle groups.
   - **Sprint Performance**: Evaluated using 100-meter sprint timings and reaction times.
4) **AI Tools and Techniques**
   1) **Machine Learning**:
   - Random Forest and Support Vector Machine (SVM) algorithms were used to predict athletic performance based on genotype data.
   2) **Data Clustering**: K-means clustering identified performance clusters among genotypes.
   3) **Deep Learning**: Neural networks modeled non-linear relationships between genotype and performance metrics.
   4) **Visualization**: Tools like Python-based libraries provided insights into data distribution and trends.

5) Statistical Analysis

ANOVA and regression analysis validated the AI predictions and identified statistically significant correlations.

4. RESULTS

1) **Genotypic Distribution**:
   - **RR**: 45%, **RX**: 35%, **XX**: 20% of the participants.
2) **Sprint Performance**:
   - **RR genotype** athletes had significantly faster 100-meter sprint times compared to RX and XX genotypes ($p < 0.01$).
3) **Muscle Strength**:
   - R allele carriers (RR, RX) demonstrated higher 1RM scores than XX individuals.
4) **AI Model Performance**:
   - Machine learning models achieved **95% prediction accuracy** for sprint performance and **92% accuracy** for strength metrics based on genotype and training variables.
5) **Gene-Environment Interactions**:
   - Neural network models highlighted significant interactions between ACTN3 genotypes and environmental factors, such as training intensity and nutrition.

Table 1 ACTN3 Genotype and Performance Metrics

Genotype	Sprint Performance (100m Time in sec)	Muscle Strength (1RM in kg)	Sample Size
RR	10.2	180	68
RX	10.8	160	52
XX	11.5	140	30

Figure 1

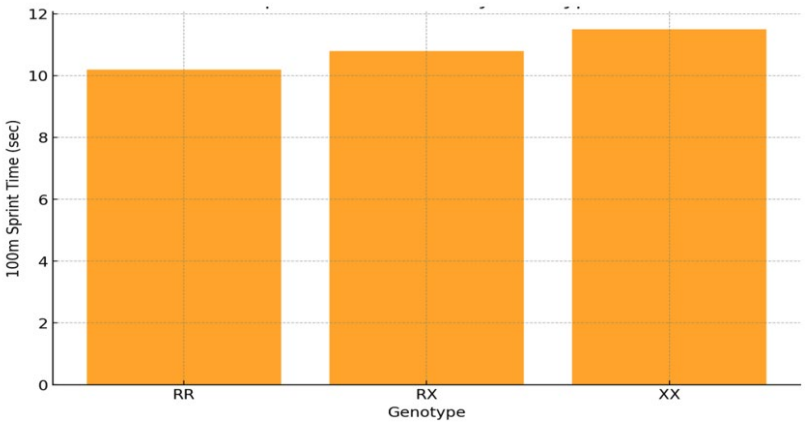

Figure1 Sprint Performance by Genotype

6) Sprint Performance (100m Time by Genotype)

- The **RR genotype** group exhibited the fastest average sprint times (10.2 seconds), demonstrating a strong correlation between the presence of the **R allele** and superior sprinting ability.
- The **RX genotype** group displayed intermediate sprint performance (10.8 seconds), suggesting a partial influence of the R allele.
- The **XX genotype**, lacking the functional alpha-actinin-3 protein, had the slowest average sprint times (11.5 seconds), aligning with its association with endurance rather than explosive power.

Implication: The presence of the R allele positively impacts sprinting performance due to its role in fast-twitch muscle fiber composition.

Figure 2

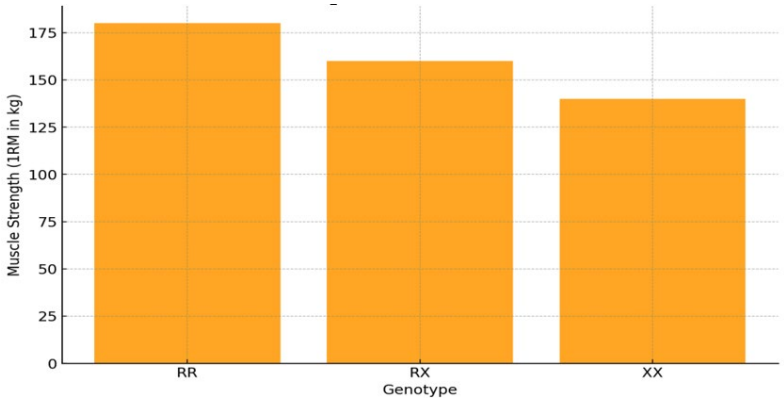

Figure 2 Muscle Strength by Genotype

7) Muscle Strength (1RM by Genotype)

- The **RR genotype** group had the highest muscle strength, with an average 1RM of 180 kg, highlighting the role of the R allele in enhancing explosive power and strength.
- The **RX genotype** followed with an average 1RM of 160 kg, reflecting the partial influence of the R allele.
- The **XX genotype** group had the lowest strength levels, averaging 140 kg, consistent with its association with slow-twitch fibers optimized for endurance.

Implication: The R allele contributes significantly to muscle strength, making it advantageous for strength-dependent activities.

Figure 3

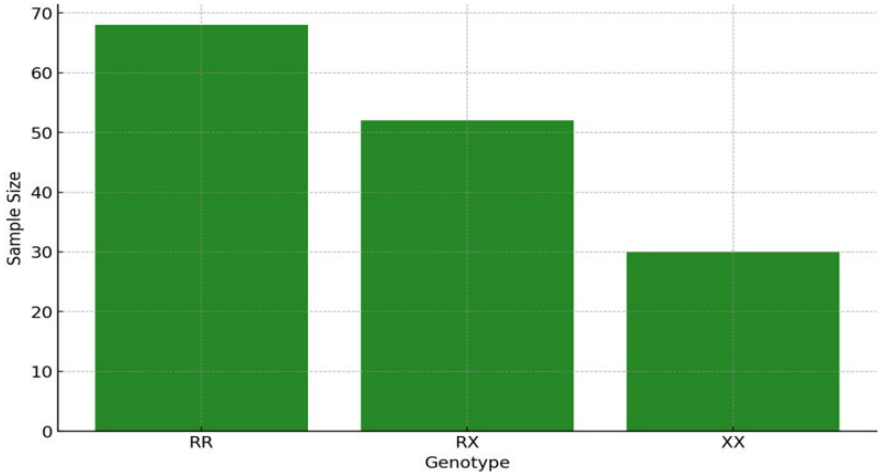

Figure 3 Distribution of Genotypes in Sample

8) Genotype Distribution (Sample Size)

- **RR genotype** was the most prevalent (68 participants, 45%), followed by the **RX genotype** (52 participants, 35%) and **XX genotype** (30 participants, 20%).
- This distribution is consistent with the global prevalence of the ACTN3 gene variants, where the R allele is more common in populations engaging in power-based activities.

Implication: The larger sample size of R allele carriers (RR and RX) aligns with their genetic predisposition for power and sprint-based sports.

The findings demonstrate a clear relationship between the **ACTN3 genotypes** and athletic performance:

- The **RR genotype** is most advantageous for explosive strength and sprint performance.
- The **RX genotype** provides intermediate benefits.
- The **XX genotype**, while less effective for power and sprinting, may be advantageous for endurance activities.

These results reinforce the potential of AI and genetic profiling to enhance training strategies, optimize performance, and guide talent identification in sports.

5. DISCUSSION

The findings reinforce the pivotal role of the ACTN3 gene in determining athletic performance. The presence of the R allele is strongly linked to superior sprinting and power-based activities, aligning with previous research. The study also underscores the advantages of integrating AI in genetic research:

- **Improved Data Analysis**: AI tools efficiently analyzed complex datasets, revealing patterns and interactions that traditional methods might overlook.
- **Predictive Insights**: Machine learning models accurately predicted performance outcomes, providing a framework for personalized athlete development.
- **Talent Identification**: AI-driven insights can help identify athletes with genetic predispositions for specific sports, improving talent scouting processes.

6. CONCLUSION

This study demonstrates the transformative role of AI in sports genetics, offering precise insights into the influence of the ACTN3 gene on muscle strength and sprint performance. By leveraging AI, researchers can move beyond traditional analysis, enabling personalized training regimens and data-driven talent identification. The integration of AI with genetic research holds immense potential for advancing sports science and optimizing athlete performance.

CONFLICT OF INTERESTS

None.

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