DOCUMENT SUMMARY This genetic research study provides powerful evidence that common risk alleles for Autism Spectrum Disorder (ASD) show widespread signatures of positive evolutionary selection. The authors hypothesize this is because these same genes are linked to enhanced cognitive abilities, neurogenesis, and brain development. This directly supports the Enlitens view that neurodivergence is an evolutionary advantage, reframing ASD not as a simple defect but as a trait with beneficial aspects that has been actively selected for in human history.

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FORMATTED CONTENT

Widespread signatures of positive selection in common risk alleles associated to autism spectrum disorder

Why This Matters to Enlitens

This paper provides foundational scientific evidence for our core belief that neurodivergent traits are evolutionary advantages, not pathologies. It uses complex genetic analysis to argue that the very genes that increase the likelihood of an autism diagnosis have been actively

selected for during human evolution because of their connection to higher intelligence and brain development.

This directly challenges the medical model's view of autism as purely a "disorder." It allows us to reframe the conversation with clients, parents, and other professionals, grounding our neurodiversity-affirming stance in evolutionary science. The paper explains the prevalence of autism not as a failure of nature, but as the "evolutionary cost" of selecting for enhanced cognitive skills in the human population—a powerful concept for our whitepapers, assessments, and advocacy.

Critical Statistics for Our Work

- **Correlation with Positive Selection**: Autism Spectrum Disorder (ASD) GWAS results are positively correlated with signatures of incomplete positive selection (p=3.53∗10−4).
- **Increased Probability of Selection**: Genetic variants associated with ASD (GWAS p<0.1) have a 19% increased probability of being in the top 5% of genomic regions showing positive selection (Odds Ratio = 1.19, p=9.56∗10−7).
- **Genetic Correlation with Intelligence/Education**: ASD genetics shows a strong positive correlation with several advantageous traits:
  - Years of schooling (rg=0.277,p=2.9∗10−13).
  - College completion (rg=0.339,p=1∗10−6).
  - Childhood intelligence (rg=0.425,p=5.74∗10−5).
- **ASD Prevalence**: A 2012 CDC report noted an ASD prevalence of 1.47% in the US population (1 in 68 children).
- **Heritability of ASD**: Common genetic variation explains 49% of ASD heritability, while inherited rare and de novo mutations account for only 6%.

Methodology We Can Learn From

The study used a purely computational and statistical methodology, which, while not directly replicable in our clinical practice, provides the basis for its powerful conclusions. The authors analyzed large-scale Genome-Wide Association Study (GWAS) summary statistics from the Psychiatric Genomics Consortium for five psychiatric conditions, including ASD.

They used a machine-learning algorithm called Hierarchical Boosting (HB) to identify genomic regions that showed signatures of positive selection in a European population dataset. By cross-referencing the locations of ASD-associated genetic variants with these signatures of selection, they were able to test the hypothesis that ASD risk alleles have been evolutionarily advantageous. This approach of combining large datasets to uncover patterns that challenge existing paradigms is philosophically aligned with the Enlitens mission.

Findings That Challenge the System

The study's primary finding is a direct challenge to the simple "disorder" model of autism. It suggests a dual mechanism for autism genetics that explains why it persists at a relatively high rate in the population.

1. **Rare, Damaging Mutations are Selected Against**: The paper acknowledges that rare, disruptive genetic mutations that can cause autism are under "purifying selection," meaning evolution actively tries to remove them. This aligns with the traditional view of a genetic disorder.
2. **Common, Risk Alleles are Selected For**: In contrast, the much more common polygenic variants that contribute to autism risk are under *positive* selection. These are not "bad" genes; they are genes that, on average, confer an advantage, likely related to cognitive ability. An individual developing autism can be seen as the "evolutionary cost" of a species-level adaptation for higher intelligence.

This complex picture shatters the black-and-white view of "autism genes" as being defective. It suggests that the genetic liability for autism is intricately linked with the genetic architecture of uniquely human cognitive strengths.

Populations Discussed

The genetic analysis was conducted using GWAS summary statistics and selection scores calculated for a

European (CEU) population from the 1,000 Genomes Project. The findings are therefore most directly applicable to individuals of European ancestry.

Alternative Approaches Mentioned

This entire paper represents an alternative framework for understanding the persistence of autism. Rather than viewing it solely through a medical/pathological lens, it applies the principles of evolutionary biology and population genetics.

- **Polygenic Adaptation**: The study frames ASD as a potential outcome of "polygenic adaptation," where subtle changes in the frequency of many different genes, each with a small effect, combine to produce an adaptive trait (like higher cognition) in the population.
- **Evolutionary Cost Hypothesis**: The paper's conclusion—that autism's prevalence is an evolutionary trade-off for enhanced cognitive abilities in the human species—is a powerful alternative explanation that moves beyond simple deficit-based models.

Quotes We Might Use

- **On the core hypothesis**: "Accordingly, we hypothesize that certain ASD risk alleles were under positive selection during human evolution due to their involvement in neurogenesis and cognitive ability."
- **On the dual nature of ASD genetics**: "at least two different evolutionary mechanisms appear to be present in relation to ASD genetics: 1) rare disruptive alleles eliminated by purifying selection; 2) common alleles selected for their beneficial effects on cognitive skills."
- **On explaining ASD prevalence**: "This scenario would explain ASD prevalence, which is higher than that expected for a trait under purifying selection, as the evolutionary cost of polygenic adaptation related to cognitive ability."
- **Connecting autism to high intelligence**: "multiple forms of evidence support that autism and high intelligence quotient share a diverse set of correlates, such as large brain size, fast brain growth, increased sensory and visual-spatial abilities, enhanced synaptic functions, increased attentional focus, high socioeconomic status, more deliberative decision-making, and high levels of positive assortative mating"
- **On the role of common variants**: "Conversely, genetic predisposition to ASD due to common variants is highly polygenic and, taken together on a population level, these alleles present beneficial effects with respect to cognitive ability."
- **The overall conclusion**: "In conclusion, the present study provides evidence regarding the role of human evolution in shaping the genetic architecture of psychiatry disorders,

providing a hypothesis to explain the ASD prevalence as the evolutionary cost of the polygenic adaptation of the disease risk alleles."

Clinical Implications

The clinical implications of this research are primarily in how we frame and understand autism.

1. **Reframing for Clients and Parents**: This study provides a scientific basis for a strengths-based perspective. We can explain that the genetic factors contributing to autism are not just "defects" but are linked to evolutionary advantages in human cognition. This can reduce stigma and self-blame, fostering a more positive identity.
2. **Explaining Prevalence**: The "evolutionary cost" hypothesis gives a coherent, non-pathologizing explanation for why autism exists. It helps answer the "why" question in a way that is empowering, suggesting that the traits are a natural and expected part of human diversity that has been beneficial to our species as a whole.
3. **Connecting to Strengths**: The documented genetic correlation between ASD and traits like childhood intelligence, college completion, and years of schooling provides strong evidence to support our focus on identifying and building upon the cognitive strengths of our autistic clients. It shows that the connection between autism and high ability is not just anecdotal but has a biological basis.