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Variability in response to traumatic injury considering genetics and pathophysiology of the disease

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Abstract
Trauma is one of the most common causes of death all over the world. The concept of trauma is very heterogeneous, due to its causes, types of injuries, severity, individual genetic variations and pathophysiological variability. This review explores the influence of these factors on trauma response and their forensic implications.
This work synthesizes and analyzes scientific literature on the influence of genetic factors on the response of fatal traumatic injuries, concerning the interplay between genetics, trauma pathophysiology, and forensic interpretation. Trauma mechanisms include blunt, sharp, penetrating, and combined forces, each producing distinctive injury patterns. Pathophysiological aspects such as inflammatory response, haemorrhagic shock, multiple organ dysfunction syndrome, and the influence of pre-existing diseases contribute to the complexity in the forensic analysis of trauma-related deaths. Cases where trauma interacts with pre-existing disease such as coronary artery disease, aneurysms, pulmonary embolism, or genetic disorders like osteogenesis imperfecta are also medico-legal relevant. Genetic variability modulates individual response to trauma, influencing outcomes such as recovery, disability or death. Single nucleotide polymorphisms in genes such as APOE, IL-1, IL-6, TNFα, COMT, BDNF, NOS3, and ACE have an influence in traumatic brain injury response.
Recognising genetic and physiological variability enhances forensic interpretation of trauma-related deaths, improves accuracy in cause-of-death determination, and clarifies medico-legal accountability. Although most research focuses on traumatic brain injury (TBI), expanding investigations to other trauma types is essential.

Keywords: Genetics, traumatic injury, pathophysiology, response to injury, legal medicine

Introduction
Trauma is one of the most common causes of death all over the world. Even though, there is not enough statistical data, since this issue has not been sufficiently investigated, specifically in Europe [17]. Although the USA is the country that presents more numbers in this matter, this study will be focused in Europe, as long as the research makes it possible to. In Portugal, there are very few studies regarding this topic.
Trauma refers to any injury to living tissue caused by an extrinsic agent. The classification of traumatic injuries is based on the mechanism of injury and the appearance of the wound.
Mechanisms that can produce injuries are classified as blunt force mechanisms, sharp force mechanisms, penetrating mechanisms and combined injury mechanisms. The application of a blunt force, delivered through a rigid object with a flat surface, results in considerable mechanical energy on the skin. Typically, such impacts can induce trauma injuries that can be caracterized as abrasion, bruise and laceration. Sharp force trauma is characterized by a cutting mechanism involving the application of pressure combined with sliding motion of a bladed instrument across the body surface, resulting in a disruption of the skin's structural integrity. Perforating trauma arises from the application of a substantial force concentrated on a reduced surface area, resulting in a wound tract that is deeper than its surface dimension. Combined injury mechanisms encompass a spectrum of traumatic injuries, including sharp-penetrating, sharp-blunt and perforating-blunt mechanisms. Other types of traumatic mechanisms that may occur, although less frequently, include asphyxia, thermal trauma, chemical trauma and electrical trauma.

The concept of trauma is very broad and heterogeneous. Therefore, there is a wide range of trauma, as well as in associated individual response and in mortality rates associated with it [14]. Traumatic brain injury TBI is one of the most fatal forms of trauma, responsible for a large proportion of deaths, for this reason is one of the most studied forms of trauma. The complexity of TBI is further exacerbated by the heterogeneity in patient outcomes, even among similar injuries, ages and health statuses [4].

The aim of this study is to critically review and analyze scientific literature on the influence of genetic factors on the response of fatal traumatic injuries. To do so articles that contain information on key biomarkers, genetic polymorphisms, as well as pathophysiological mechanisms involved in trauma response in the light of the forensic field were reviewed.

A comprehensive understanding of this matter will potentially contribute to the development of more effective investigations and develop this forensic field.

Traumatic brain injury-TBI

Traumatic Brain Injury (TBI) involves damage to the central nervous system resulting from external mechanical force and can be categorized through various criteria, including etiology, severity, the extent of brain tissue involvement and additional factors. [16] It is characterized by a complex pathophysiology that unfolds in two distinct phases: primary and secondary.

The primary phase involves mechanical forces disrupting the brain parenchyma and blood-brain barrier (BBB) at the moment of impact. The secondary phase involves a systemic and neuroinflammatory response mediated by immune cells and the activation of neural cells, triggering the release of cytokines, growth factors and adhesion molecules. This secondary injury can evolve over hours, days, or even months after the initial trauma, potentially leading to metabolic dysregulation and further brain injury [2]. TBI results in acute alterations to the brain's cytoarchitecture, leading to diffuse axonal injury, dendritic shearing, synaptic disruption and demyelination. These effects may be exacerbated by hematoma-induced pressure, loss of white matter and additional injury mechanisms that manifest in later phases, including necrosis, apoptosis and ongoing inflammation [16].

The outcomes following TBI can be variable, even among similar injuries. The aim of some studies is to verify whether the outcomes are consistent with the importance of genetics in the response to trauma, particularly in cases of TBI.

Inflammatory response to trauma

Traumatic Haemorrhagic Shock (HS) is a severe consequence stemming from injury that greatly adds to the total number of traumatic deaths while also dramatically complicating outcomes, specifically after a Traumatic Brain Injury (TBI). Together HS and TBI account for roughly half of all deaths from trauma within the first 24 hours of hospital admission [8]. The combination of substantial haemorrhage and significant trauma sets off a series of post-traumatic physiological changes, inflammatory changes, and immune system reactions that can lead to damage or death of multiple organ systems. Mechanistically, systemic inflammation and organ injury in traumatic HS involve coagulation, the complement system, impaired microcirculation, and inflammatory signalling pathways [8]. Organs such as the liver, spleen, or sometimes the lung may be injured within their capsules following trauma, even if the covering membranes remain intact. Renal failure is a common sequel to trauma; destruction of the renal tubular epithelium can occur in cases of crushed muscle, extensive burns, or exposure to toxins like mercuric salts. Trauma can also compromise arterial or venous walls, leading to false aneurysms that may later rupture, or the formation of arteriovenous fistulas that can burst [16]. Multiple Organ Dysfunction Syndrome (MODS) is identified as one of the leading causes of late death after traumatic injury, as it was mentioned earlier, representing a state where organ injuries complicate the primary trauma damage. MODS is partly attributed to excessive or maladaptive activation of inflammatory pathways [8]. Infections at trauma sites can involve adjacent vessels, potentially causing severe haemorrhage if the vessel wall is eroded by an abscess or cellulitis. Sepsis, defined as an "unusual systemic reaction" to infection, involves a biphasic immune response with hyper-inflammatory and immunosuppressive phases, often leading to multiple organ dysfunction and susceptibility to nosocomial infections [16]. Severe sepsis occurs when sepsis is accompanied by evidence of organ dysfunction, potentially progressing to septic shock with cardiovascular collapse, often refractory to treatment [9]. The organ failure seen in severe sepsis is similar to MODS in patients who survive severe traumatic injuries, with studies confirming a link between systemic inflammation and Multiple Organ Failure (MOF) [8, 9].

Disease and trauma

In forensic pathology, one of the most complex and contentious issues arises when a death occurs in an individual who has suffered trauma but who also has a pre-existing natural disease or where a natural disease emerges after trauma.

Three hypotheses must be addressed:

- Whether death was caused solely by the natural disease, independent of any trauma.
- Whether death was caused entirely by the trauma, irrespective of the pre-existing pathology.
- Whether death resulted from a synergistic interaction between the trauma and the natural disease.

The considerations mentioned above are especially pertinent in cases involving coronary artery disease, subarachnoid haemorrhage and pulmonary embolism [16].

Osteogenesis Imperfecta (OI) is a bone disease and a heritable disorder of connective tissue. It is caused by a polymorphism is genes COL1A1 and COL1A2 that encode type I collagen. The relationship between these gene variants and trauma is direct and profound, primarily manifesting as bone fragility and a high susceptibility to fractures. Individuals with OI often sustain fractures with minimal or no trauma, affecting mostly bones in extremities [5, 15].

The forensic assessment of deaths involving both natural disease and trauma requires a nuanced, objective, and evidence-based approach, according to the autopsy findings and clinical history. Determining whether death was a direct consequence of trauma, an inevitable outcome of disease, or

the result of their interplay lies at the heart of forensic pathology.

Genetic polymorphisms in trauma response

The human organism consists of a complex multicellular system in which cellular functions are governed by a genome made up of DNA. Genetics encompasses the study of heredity and variability in organisms. Within the genome, composed by nucleotides, polymorphisms can occur, which are natural variations in the DNA sequence. Single nucleotide polymorphisms (SNPs) are one of the classes of polymorphisms that can occur, which constitute common genetic variants, often associated with an increased risk of disease.

Models that study SNP polymorphisms have been utilized to examine the diverse responses of individuals to injury and other inflammatory or infectious stimuli. It is important to note that while SNPs themselves do not originate disease, it is possible they can to affect the risk of developing a disease or the severity of a disease or condition. Concerning traumatic injury, the host initiates a cascade of biological processes aimed to mitigating damage and promoting repair. Individual's genetic, specifically the presence of relevant polymorphisms, can modulate these responses.

Research on the genetic factors influencing recovery from traumatic injuries has been predominantly focused on traumatic brain injury (TBI), with limited data available for other traumatic injuries in general. Consequently, the following discussion is focused on studies and findings specific to TBI [2, 11].

Materials and Methods

A comprehensive search of relevant and reliable databases, including Google Scholar and PubMed. Initial screening was based on abstracts and conclusions, focusing on studies that explicitly examine the association between genetic variations and response to trauma. Subsequently, full text articles were evaluated regarding the methodologies used and their relevance to the research questions. More studies focused on traumatic brain injury (TBI) were conducted than those that examined general trauma. Consequently, the influence of genetic factors on TBI was also more frequently investigated.

The data extraction aimed to summarize the main findings, including specific genes, types of traumatic injuries studied, and the outcome measures used. The synthesis of the extracted data focused on identifying consistent patterns and discrepancies among studies. By synthesizing the existing literature, this review aims to provide a comprehensive overview of the current understanding of the genetic basis of variable responses to traumatic injury.

Results and Discussion

Inflamatory Markers, Cytocines, proteins and other regulators and TBI

As previously mentioned, genetic research related to traumatic injuries has not been covering all types of injuries, with most studies and specific findings focusing on TBI.

Among the most studied inflammatory markers, cytokines and their genetic polymorphisms have been linked to differences in clinical outcomes following TBI.

The tumour necrosis factor alpha (TNFα), a cytokine that regulates neuronal death and synaptic plasticity, has a well-known polymorphism-the TNFα-308 a allele-which is associated with poorer outcomes due to elevated expression of this pro-inflammatory molecule. This polymorphism translates in a higher expression of TNFα and, consequently, a higher possibility to severe disability and unfavourable outcomes in TBI survivors. In different analysed studies, it also suggests that it did not impact significantly in mortality [2, 4].

Similarly, interleukin-1 (IL-1), particularly its IL-1α and IL-1β isoforms, plays a key role in amplifying inflammation and inducing fever, following injury. The IL-1β + 3953 T allele has been correlated with increased intracranial pressure and greater injury severity in TBI patients. [10]

Polymorphisms in the IL1RN gene have also been target of study in relation to TBI. IL1RN2 allele has been associated with higher rates of haemorrhagic contusions, which results in less favourable outcomes following TBI. [3]

Interleukin-6 (IL-6) production is stimulated by TNFα and IL-1. It represents another cytokine with dual effects: while it can exert neuroprotective actions, it also contributes to neurotoxicity under certain conditions. The IL-6-174 GG genotype has been linked to more favourable outcomes post-injury and could be associated with a lower risk of death following TBI. Whereas individuals with the IL-6-572 CC genotype might exhibit increased susceptibility to concussion [2, 12].

Apolipoprotein E (APOE) is, to date, the most extensively studied protein in the context of TBI. It plays a pivotal role in neuronal repair, growth and modulating the inflammatory response. E2, E3 and E4 are three common isoforms, but the APOE4 allele is of particularly interest, which has been consistently associated with worse clinical outcomes, including prolonged hospital stays, greater injury severity and an increased risk of developing post-traumatic Alzheimer's disease. Some studies show that individuals with an APOE4 allele were less tolerant to increases in cerebral perforation pressure which can be related to mortality risk and it was also linked to higher fatality rates [2, 4, 6].

Transforming growth factor beta (TGFβ), a regulator of inflammation and tissue repair, during the acute phase of TBI, due to the inhibition of IL-1, IL-6 and TNFα production. The TGFβ-509 T allele may confer greater neuroprotection in the outcome of TBI. [2]

Beyond cytokines and APOE, several other genetic polymorphisms have demonstrated relevance in TBI prognosis. For instance, the tau protein (MAPT), that is involved in stabilizing microtubules, when hyperphosphorylated, is a hallmark of various neurodegenerative disorders such as Alzheimer's disease and chronic traumatic encephalopathy (CTE). Following TBI, elevated MAPT levels have been detected in cerebrospinal fluid and plasma, which corelates it with the severity of the injury and the outcome. Additionally, the Tau Ser53Pro TT variant may enhance the risk of recurrent concussions [2].

The NOS3 gene, encoding endothelial nitric oxide synthase, is essential for regulating cerebral blood flow. The -786 C allele of NOS3 has been associated with reduced perfusion following TBI, potentially worsening outcomes [2, 11].

The catechol-O-Methyltransferase (COMT) gene, which modulates dopamine levels in the prefrontal cortex, also plays a role in cognitive outcomes after TBI. The Val158Met polymorphism is linked to high enzyme activity

and lower dopamine levels. It appears to impact executive functioning, with Val allele carriers showing poorer performance, following TBI [2, 4, 11].

Additionally, the angiotensin-converting enzyme (ACE), known for its role in blood pressure and cerebral perfusion regulation, presents a D/D genotype that has been linked to worse cognitive outcomes post-injury [2].

Brain-Derived Neurotrophic Factor (BDNF), a key modulator of synaptic plasticity and memory, is also relevant; individuals carrying the Val66Met polymorphism may experience reduced cognitive performance following TBI [2, 4, 11].

Furthermore, tumour protein p53 (TP53), a critical regulator of apoptosis, is implicated in injury outcomes as well. The Arg/Arg genotype of TP53 has been associated with more severe prognoses in cases of major TBI. Studies don't state a direct correlation with mortality after TBI, but it states a bad outcome, which can be related with higher mortality [2, 11].

Mitochondrial genetics and TBI

Lastly, mitochondrial genetics is an emerging field in TBI research. Given the mitochondria's central role in energy production and oxidative stress regulation, certain mitochondrial DNA polymorphisms have shown prognostic potential. Specifically, the mtDNA haplogroup K has been linked to better recovery outcomes, potentially lessening the negative effect of aging. While the A10398G variant appears to influence recovery trajectories in a sex-specific manner [2].

Other important genes that may influence trauma outcomes

Regarding trauma in general, COL1A1 and COL1A2 genes are responsible for encoding the alpha-1 and alpha-2 chains of collagen type I, respectively. Collagen it's a very important protein present in bones and other tissues. Abnormalities in these genes make the individual more prone to fractures in bones and progressive deformity, which can increase risk of mortality [15].

The following table presents these findings in a more summarized format, where protective suggest a protective effect against severe trauma outcomes and risk factor suggest a negative effect, increasing risk of complications and even death.

Table 1: Studied polymorphisms and its characteristics

Gene / Polymorphism	Main Function / Mechanism	Impact on Trauma Outcome	Interpretation
IL-6-174 GG	Regulates inflammation; anti-inflammatory profile	Linked to better neurological recovery	Protective
TGF-β-509 T	Inhibits pro-inflammatory cytokines (IL-1, IL-6, TNF-α)	Promotes neuroprotection and healing	Protective
mtDNA Haplogroup K	Enhances mitochondrial resilience to oxidative stress	Associated with improved recovery after TBI	Protective
TNF-α-308 A	Increases TNF-α production; promotes inflammation	Linked to worse neurological outcomes	Risk factor
IL-1β +3953 T	Amplifies inflammatory response	Associated with increased intracranial pressure	Risk factor
*IL1RN*2*	Alters regulation of IL-1	Correlated with haemorrhagic brain contusions	Risk factor
IL-6-572 CC	Modulates inflammatory profile	Linked to higher susceptibility to concussion	Risk factor
APOE ε4	Modulates neural repair and lipid transport	Associated with poor recovery, post-traumatic dementia	Risk factor
MAPT Ser53Pro TT	Involved in microtubule stability	Increases risk of chronic traumatic encephalopathy	Risk factor
NOS3-786 C	Reduces nitric oxide production; affects cerebral blood flow	Linked to worsened perfusion and outcome	Risk factor
COMT Val158Met (Val/Val)	Affects dopamine breakdown and cognitive function	Associated with poorer executive functioning post-TBI	Risk factor
ACE D/D	Increases vascular tone and resistance	Correlated with poor cerebral perfusion	Risk factor
TP53 Arg/Arg	Regulates apoptosis and DNA repair	Related to poor prognosis in severe TBI	Risk factor
COL1A1 / COL1A2 (mutations)	Encodes type I collagen; maintains bone and vessel integrity	Increases risk of fracture and vascular failure	Risk factor

These findings emphasise the relevance of considering genetic susceptibility when interpreting traumatic deaths, highlighting the potential benefits of incorporating these molecular profiling into forensic assessments.

As already mentioned, molecular profiling would help to improve the understanding on how individual variations can influence in trauma response and mortality risk among individuals. A possible approach would involve establishing a biobank with relevant biological samples, such as blood, tissues, ensuring ethical and legal frameworks [1, 16]. Using next-generation sequencing (NGS) for targeted genotyping could provide relevant variants, even more if complemented with other techniques like quantitative PCR or Sanger sequencing for further validation [3-6, 10, 11]. Additionally, using techniques like RT-qPCR and ELISA would tie each genetic variant to actual gene activity and cytokine levels, revealing how trauma related molecular mechanisms operates [5, 8, 12]. Ultimately, combining, these molecular approaches with ethical and legal frameworks concerning the treatment of genetic data, could promote the establishment of validated forensic biomarkers and encourage the establishment of national and international trauma-specific genetic databases [1, 16].

Conclusion

A comprehensive review of the existing literature reveals that understanding trauma-related mortality is incomplete

without considering the biological variability of each individual.

The interaction between inflammatory response, haemorrhagic shock and multi organ failure it an important factor in traumatic death. The duration, severity and anatomical location of injury are crucial for the risk of mortality, and these effects are worsened by genetic and immunological factors.

It is important to note that these mechanisms are shaped by genetic polymorphisms that influences immune regulation, coagulation pathways, neuroprotection and metabolic responses. The presence or absence of specific alleles has been shown to significantly alter the course of recovery or fatality. Consequently, trauma should be assessed not only in terms of its physical injury, but also as a biologically modulated event with deeply individualised outcomes.

Another crucial challenge is to distinguish the contributions of trauma and pre-existing disease to a fatal outcome. Cases involving coronary artery disease, subarachnoid haemorrhage, pulmonary embolism or osteogenesis imperfecta highlight the need for a contextual approach that goes beyond simplistic causal hypotheses.

Nevertheless, there are significant limitations in this research. There remains low investment in the field, a lack of large-scale forensic genetic studies in humans, an absence of standardised protocols and limited access to advanced testing techniques. These barriers emphasis the need for ongoing research and substantial investment.

In future perspectives, research should expand genomic databases with trauma specific data, the validation of forensic biomarkers in diverse populations and further investigations related to trauma, even for clinical subjects, regarding trauma prevention and response, in order to reduce traumatic mortality rates.

As a final conclusion, this study validates the need to understand the complex nature of the subject, which includes external injuries, internal conditions, genetic factors and complex biological events involved in trauma outcomes. The future of legal medicine and forensic pathology relies in its capacity to combine these aspects, with the aim of objectively explaining the manner of death and its underlying causes.

Table 2: List of abbreviations

Abbreviation	Meaning
TBI	Traumatic brain injury
GCS	Glasgow Coma Score
CNS	Central nervous system
MOF	Multiple organ failure
ISS	Injury Severity Scores
SAH	Subarachnoid haemorrhage
HS	Heamorrhagic shock
MODS	Multiple organ dysfunction syndrome
DVT	Deep vein thrombosis
OI	Osteogenesis imperfecta
DNA	Deoxyriobonucleic acid
SNP	Single nucleotide polymorphism
PCR	Polymerase chain reaction
RT-qPCR	Quantitative reverse transcription polymerase chain reaction
ELISA	Enzyme-Linked Immunosorbent Assay

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