CASE REPORT

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Cause of an Aberrant Phenotype: An Interesting Dilemma

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ABSTRACT: In a recent homicide case, the discovery of an unusual group specific component (Gc) pattern in a bloodstain provided compelling evidence linking the suspect to the scene of the crime. The phenotype could not be identified, nor could the source of the aberrant pattern be attributed to either the victim's parents. Some possible explanations are presented.

KEYWORDS: pathology and biology, genetic typing, human identification

Late in the evening of 13 Dec. 1979, the partially clothed body of a young female was found along the shoulder of a rural road. The body was mutilated by multiple stab wounds to the neck that had severed the carotid artery and by overlapping lacerations extending from the pubis to the sternum.

At the crime scene, a partial bloody track from a shoe or boot was found on the road near a pool of blood adjacent to the victim's body. The track and samples of blood from the pool were the only items of physical evidence collected at the crime scene.

A suspect was subsequently identified, and fragmentary bloody sole tracks were located on the floor mat of his pickup truck. The sole pattern of the suspect's boots was similar in class characteristics to the bloody impressions left at the crime scene and on the truck's floor mat.

Examination of the soles of the boots failed to locate visible bloodstains; however, processing of the boots with luminol [*1*] revealed probable blood traces on the sole of the right boot. It was later learned that the suspect had "washed" his boots in the detention cell sink.

Analyses of the bloodstains from the floor mat and crime scene and of the specimens from the victim and suspect were conducted for a variety of blood group polymorphisms. Not only did this work reveal a consistency between the victim's phenotypes and those identified in the bloodstains, but a particularly interesting protein pattern was also noted in the group

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specific component (Gc). Conventional electrophoretic analysis was employed with cellulose acetate membranes as the support media [*2,3*].

The electrophoretogram revealed a four-banded pattern unlike the common Gc type 1-1, 2-2, or 2-1 patterns (Fig. 1). A review of the available literature failed to identify a similar pattern. The question then arose as to whether the expressed Gc pattern was a true genetic variant or an anomaly resulting from alteration of the original proteins.

Because of the possibility of a common Gc pattern changing to the observed pattern by degradation processes, the following facts regarding the various specimens were considered:

1. The blood specimens from the pool of blood and the victim gave identical electrophoretic patterns.
2. The cool evening temperature was nearly ideal for preservation of Gc patterns at the time the crime specimens were deposited and collected.
3. The body had cooled rapidly because of the cold day's air, and the autopsy was performed within 9 to 10 h of death.
4. Neither the crime scene nor the autopsy specimen was contaminated with other body fluids such as vomitus, bile, or urine resulting from trauma.

Discussion

The four-banded pattern proved to be stable and reproducible; repeated analyses of both stained and fresh blood specimens consistently yielded the same electrophoretic patterns.

The pattern has four bands, the three most anodal bands having the same relative intensities and interband spacings as three type 2-1 bands (Fig. 1). However, the entire variant pattern appears shifted slightly more than the type 2-1 patterns toward the anode. The

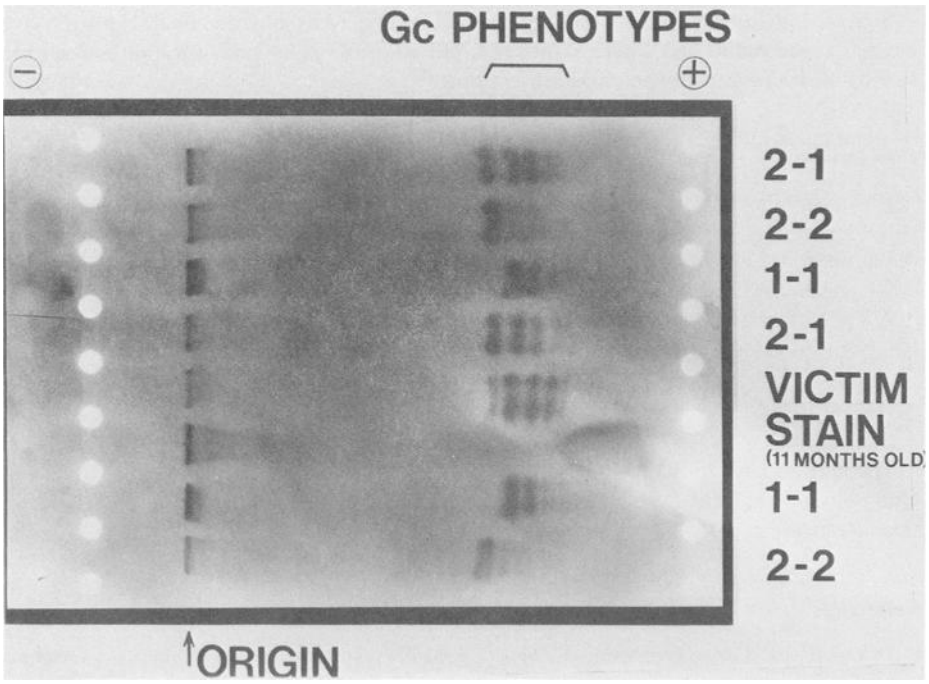

FIG. 1—*The electrophoretogram illustrates the unusual pattern exhibited by the victim in relation to the three common Gc phenotypes.*

fourth band of this pattern is much less intense but distinct, and it migrates cathodally to the other three.

Comparison of this pattern with a previously encountered four-banded pattern, discovered by Grunbaum [*4*], revealed distinct differences in the electrophoretic mobility and relative band intensities.

In an attempt to discover the source of the four-banded pattern, blood samples were collected from the victim's parents and three siblings. It was disappointing not to find the same pattern in any of the family members tested. The mother and father were Gc types 2-2 and 1-1, respectively. However, comparison in other polymorphisms was in agreement with expected distributions.

As the victim is deceased and not available for continued study and the source not identified with the parents, we can only speculate as to the cause of the observed pattern. Some possible explanations are these:

1. A mutation at the Gc locus occurred independent of each parent.
2. A mutation occurred in the maternal gamete and the putative father is not the biologic father.
3. The victim was not the biologic child of either putative parent, perhaps through a maternity ward mix-up.
4. An unusual combination of post-translational modifications of the allele expressions of Gc^1 and Gc^2 may have taken place [*5*]: (a) The victim was actually a Gc 2-1. The Gc^2 allele may show a two-banded pattern, as occasionally happens, or the anodic component of the Gc allele may have split to produce two visible bands, thus yielding a multibanded pattern. The distribution of these bands may have been affected by post-translational changes such as (b) and (c). (b) Complete or partial desialylation of the carbohydrate side chain produced an unusual electrophoretic band distribution [*6*]. (c) The amount of vitamin D complexed with the Gc protein molecule affected electrophoretic mobilities and band distribution [*7*].
5. Since four-banded Gc patterns are now known to exist as inherited characteristics,[4] it is likely that the victim had a heterozygous Gc phenotype in which one of the rare alleles carried by an ancestor expressed itself at this time.

Conclusions

1. Phenotyping for Gc should be attempted in every forensic science investigation of blood and bloodstains, provided the specimen is "viable," because of its excellent frequency distribution, the great variety of rare variants, the persistence of the Gc proteins relative to other polymorphisms, and ease of determination.
2. The observation of an unusual pattern, though not identified by a classified name, can be of critical value in the resolution of legal or criminal investigations.

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