



## LETTER TO THE EDITOR

Dear Sir,

### INCONSISTENCY IN RESULTS OF ADULT HAEMOGLOBIN GENOTYPE AND ITS IMPACT ON PRENATAL DIAGNOSIS: LESSONS FROM A PRENATAL DIAGNOSIS UNIT

Prenatal diagnosis of sickle cell disorder has existed in Nigeria for about a decade, with publications on various aspects of the procedure and service and its improving utilization in spite of the limitations caused by socio economic factors.<sup>1,2,3</sup> In a unit in Nigeria, all cases of prenatal diagnosis of sickle cell disorders were through transabdominal chorionic villous sampling, following standard clinical and laboratory steps. As a prelude to performing the procedure, the women are expected to fill a questionnaire, part of which includes the known genotypes of both the husband and wife. It is known that the result of the fetal analysis is derived from that of the parents. The blood samples of the women were routinely collected as part of the standard procedure. The husband's blood was not mandatory as we relied on the results disclosed by the women. We observed inconsistencies between the results of the fetal genotype and the couple's genotypes and this prompted further enquiry about the couple's genotype on a few occasions. This letter is intended to share with other scientists our experience about inconsistencies in genotype results and to also offer advice to other prenatal diagnosis centres on the ways of avoiding similar pitfalls.

#### Case 1

Mr and Mrs A were both health professionals. She had chorionic villous sampling in the first pregnancy with the fetal genotype being AC. The parents were both of genotypes AS. The laboratory analysis was repeated thrice because of the presence of the C gene and was consistently confirmed as AC. The parents were invited for interview and

genotype confirmation and the result showed that father was AC.

#### Case 2

Mrs B, presented for prenatal diagnosis on the basis that her fetus was at risk of SS. She was gravida 4, para 3<sup>+0</sup>, 2 alive. The last baby was diagnosed as a sickler (SS), shortly before he died in a private hospital. Prior to this event, she had an AA genotype result, while the husband was confirmed to be AS by a private laboratory in the East. Hence the couple was shocked at the disclosure of the baby's genotype. All the other children are AA. The fetal genotype result was AA (repeated twice), while the couple were both AA.

#### Case 3

Mr and Mrs C are both health professionals. They presented for prenatal diagnosis of fetal genotype in the third pregnancy. The second baby was confirmed to be SS after several medical consultations to their astonishment. They had always known their genotypes to be AA and AS respectively from a private laboratory in the north. The wife was later confirmed to be AS by another laboratory in Lagos after the diagnosis in the second baby. We also confirmed the AS genotype in our centre.

#### Case 4

Mr and Mrs D, another health professional and business woman had prenatal diagnosis for fetal genotype in the second pregnancy. The first baby died after a diagnosis of SS was made. The husband and wife, had always known they were AA and AS before the event. A repeat test confirmed genotypes AS for both husband and wife. The fetus was AS.

#### Implications

Inconsistencies in genotype results could have multidimensional effects on the overall life of the family, legal, medical and psychosocial.

The genotype is one of the few medical information that are admissible in law to establish the legal identity of an individual. Example of its use is in paternity dispute and issues of heredity. Errors can therefore mislead legal opinions, with far reaching consequences. The medical implication of wrong genotype could in the management. This is more so in individuals with genotype like SC that may have been wrongly diagnosed as AS. At family/psychological level disruption of family tranquility and peace is one of the severe consequences of changing genotypes. We observed that the men were usually very aggressive and almost jumped into conclusion even while we were still trying to establish the correct genotypes. If not carefully handled, it could lead to matrimonial disharmony and divorces. The most extreme of the consequences is when an SS is delivered in the family, as a result of genotype misconception.

#### Lessons

We identified that the absence of the husband and the practice of not routinely doing the couple's genotype contributed to the difficulties that we experienced. In our socio cultural setting, it is not common place for husbands to accompany their wives to clinics. Indeed in reproductive health issues, most African men would not agree to be tested for medical problem, believing that the wife is usually the culprit. We admit that we were not insistent in requesting for the husbands genotype before cvs. In the face of the magnitude of the problems of inconsistent genotype results that we had and its implications, we are now firm about authenticating the genotype of both couple. Initially, we were requesting for the blood sample to be along brought by the women in lieu of the husband's non availability. We however realized that some husbands lived far away. We then, introduced the DNA cards, which can be posted using the normal sized official envelopes, with the drop of the husband's

blood on it. This is more acceptable and convenient for the women and the husbands, irrespective of their locations.

Prenatal diagnosis centres should cautiously accept genotype results and routinely confirm same to avoid the difficulties that we experienced. Unexpected fetal genotype results should be handled with great caution to avoid the problems that could be a fall out of it.

**Conclusion:** Our observation may actually be 'a tip of the ice bag' situation of inconsistency in genotypes results. It is a challenge to scientists to be more alert

to laboratory diagnosis to avoid misleading both the individual and the attending physicians.

#### REFERENCES

1. Akinyanju OO, Disu F, Adewole TA. Initiation of Prenatal Diagnosis of Sickle Cell Disorder in Africa. *Prenat. Diagn.* 1999; **19**: 299–304.
2. Oloyede AO Akinde JA. Evaluation of Early Complications of Chorionic Villus Sampling. *Emirates Med. Journal.* 2004; **22**: 225–230.
3. Oloyede OAO. Influence of Socio-Demographic Characteristics on the

Utilization of Chorionic Villus Sampling Service in Nigeria. *Tropical Journal of Obstetrics and Gynaecology.* Oct. 2005; **22**: 116–119.

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