eNeurologicalSci 3 (2016) 33–34

Contents lists available at ScienceDirect

eNeurologicalSci

journal homepage: http://ees.elsevier.com/ensci/

Consanguinity and rare neurological disease. A five year experience from the Korle Bu Teaching Hospital, Accra, Ghana

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ARTICLE INFO

Article history:
Received 15 September 2015
Received in revised form 8 January 2016
Accepted 21 January 2016
Available online 22 January 2016

Keywords:
Consanguinity
Ghana

ABSTRACT

Introduction: Marriage between close biological kin is not regarded as advantageous in the western world but in other parts of the world, consanguineous unions persist. Consanguineous marriage increases the birth prevalence of individuals with recessive disorders. In Accra, Ghana, consanguinity is beginning to emerge as a significant cause of rare neurological disease at the central referral hospital at Korle Bu in Ghana.
Method: Documentation of rare neurological and genetic diseases over a five year period resulting from consanguinity (2010–2015) presenting to the Department of Child Health, Korle Bu Teaching Hospital, Accra.
Results: One of the three siblings with zeroderma pigmentosum was identified as the rare De Sanctis Cacchione syndrome which has not been previously reported from West Africa. Five cases of spinal muscular atrophy including three consecutive siblings with the disease, MCAD deficiency (1), inborn errors of metabolism (1), ceroid lipoid fuscinosis (6), a case of Meckel Gruber syndrome.
Conclusion: Rare neurological disease occurs in West African communities as a result of consanguinity.

1. Introduction

Genetic disorders place significant health and economic burdens on health care systems. A consanguineous relationship is one between individuals who are second cousins or closer. The purpose of this research was to document the existence of consanguinity as a basis of rare neurological disease in the paediatric population of Ghana.

A male child presented to the Korle Bu Teaching Hospital (Department of Child Health) in January 2008 with a generalized rash of two years duration. It started at the age of three months and had evolved to show a pustular rash, hypo and hyper pigmented lesions and recurrent lip ulceration. A chronic discharge had been noted from both eyes. It emerged that the children were products of a consanguineous marriage. The parents were first cousins. The paediatric ophthalmologist made a diagnosis of a squamous cell carcinoma in situ after an excision biopsy. A neurology consult was requested because of delayed development. The classic features of Xeroderma Pigmentosum (XP) were present. The child had a clear ataxic gait. His head circumference was 44 cm, less than 0.4th centile (microcephaly). His height was 94 cm (less than 0.4th centile). The penile length was 1.75 cm (micropenis). His weight was 12 kg (less than 0.4th centile). The testes were undescended. Neurologic examination demonstrated normal tone and power but reduced reflexes. Using the schedule of Growing skills 2 to assess his overall development, he was found to be functioning at 24 months instead of 36 months. A CT scan of the brain done was normal. The EEG was however abnormal, dominated by fast waves and several isolated spikes. A few endocrine tests that the parents could afford were abnormal. Luteinizing Hormone — 0.1 Miu/ml (normal 7.6), Follicle stimulating Hormone — 0.7 (normal less than 3.0 u/L), and serum Prolactin — 342 (normal 60—390 mU/L). He had cryptorchidism. He later on developed spasticity of the lower limbs and more eyelid carcinomas. On the basis of global developmental delay, ataxia and XP, a diagnosis of De Sanctis de Cacchione syndrome was made.

1.1. Case 2

A 24 h old male baby was admitted to the neonatal unit of the hospital with multiple congenital anomalies. The neonate was noted to have a small head 31 cm (<0.4th centile) and was floppy with posteriorly rotated ears. There was a ruptured, moderately sized occipital encephalocele. Other significant anomalies documented included a posterior central cleft palate, a bell shaped thorax. Enlarged kidneys were palpated with a palpable bladder and postaxial polydactyly. An autopsy confirmed polycystic kidneys. All the features were consistent with Meckel Gruber syndrome. There was consanguinity. Both parents were second cousins and from the northern part of Ghana.

A six year old boy presented in March 2014 with seizures occurring during the mornings and more recently an episode of status epilepticus.

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http://dx.doi.org/10.1016/j.ensci.2016.01.002

The final diagnosis was Medium Chain Acyl CoA dehydrogenase deficiency after documenting hypogylcaemia with no ketonuria and recurrent seizures. The mother's paternal uncle turned out to be the father of the child.

1.2. Case 4

Over a period of five years the Child Health Department recorded thirty cases of neurodegenerative disorders and 90% were from the Volta region of Ghana. 25 out of 30 cases appeared to be Neuronal ceroid lipofuscinosis from the clinical and MRI features and notably six cases showed evidence of consanguinity. They were all second cousins.

1.3. Case 5

Spinal muscular atrophy has been recorded with children from the Northern and Ewe tribes. There was one family with three male children in a row with type 2 SMA. Five recorded cases came from consanguineous backgrounds out of 14 cases over the past four years.

2. Discussion

It is likely that the impact of consanguinity has not been fully recognized in West Africa. Consanguinity is a common indication for genetic counseling but it rarely occurs in our hospitals. The highest rates of consanguineous marriage are reported from the Middle East, North Africa and Central Asia where more than 25% of the world's population live and second cousins or closer relatives account for more than fifty percent of all marriages [1]. The mountainous terrain in the Volta region of Ghana may have led to inbreeding, generation ago in my opinion. A recessive founder can rapidly increase in frequency resulting in birth of an affected child whether the parents are known to be consanguineous or believe themselves to be non-relatives [2]. The presence of consanguinity increases the probability of an autosomal recessive condition as well as non-genetic conditions and an increased risk for developmental delay and neonatal death. The risk increases when there are multiple consanguineous marriages within the same kindred [2].

It does appear that in Ghana and other parts of West Africa there are social and economic advantages of consanguineous marriage for it to persist even with the apparent decline in the Western world. There are reduced chances of maltreatment, simplified bride premarital negotiations, greater social compatibility of the bride with her husband's family, the reduced requirement for dowry and maintenance of the family goods and monies [3].

Ghana currently has a population of 27 million.

3. Conclusion

Rare neurologic disease has been recorded in Ghana as a result of consanguinity. Genetic counseling centers should be established in the Teaching Hospitals. Ethical issues in clinical genetics should be emphasized in postgraduate training in the subregion.

Conflict of interest

The authors declare that there are no conflicts of interest.

References

[1] J. Zlotogora, Y. Hujerat, S.A. Barges, et al., The fate of 12 recessive mutations in a single village, Ann. Hum. Genet. 71 (2007) 202–208.
[2] M.J. Khoury, B.H. Cohen, E.L. Diamond, et al., Inbreeding and prereproductive mortality in the old order Amish111. direct and indirect effects of inbreeding, Am. J. Epidemiol. 125 (1987) 473–483.
[3] A.K. Sagar, A. Bittles, Consanguinity and child health, Paediatr. Child Health vol. 18 (5) (2008) 244–249.