A four month female infant presenting with primary Immunodeficiency diagnosed at autopsy: a case report

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Introduction:

Di George syndrome is a type of primary immunodeficiency which results in T cell immunologic deficit due to thymic aplasia. It is manifested by severe immunodeficiency frequently presenting in infancy. In Subsaharan Africa, there are no case reports of primary immunodeficiencies. This case illustrates the findings of a case diagnosed at autopsy and the use of immunohistochemistry to evaluate the cellular lymphocyte deficiency that is diagnostic of Di George's syndrome.

Case presentation:

The decedent was a four month old African female infant who died while undergoing treatment at a referral hospital in Nairobi, Kenya. She presented with a month's history of recurrent respiratory infections, a subsequent decline in the level of consciousness and succumbed to her illness.Her two older siblings died of similar circumstances at 3-4 months of age. At autopsy, findings of thymic aplasia, bronchopneumonia and invasive fungal infections as well as minimal perilesional inflammation were characteristic of primary immunodeficiency, specifically Di George syndrome.Microbial cultures of cerebrospinal fluid, jejunal contents, spleen and lung tissue showed drug resistant Klebsiella spp, Pseudomonas spp, Serratia spp and Escherichia coli. Immunohistochemistry of splenic tissue obtained from autopsy confirmed reduction of T lymphocytes.

Conclusion:

Although rare, primary immunodeficiencies are encountered in medical practice in Kenya, unfortunately late diagnoses limit their therapeutic options and a high index of suspicion is required as well as multidisciplinary approach to diagnosis, consisting of paediatricians, internists, pathologists, microbiologists and immunologists. Use of immunohistochemistry on histological sections of tissues derived from autopsy are important for post mortem diagnosis of Di George syndrome. This report also highlights the requirement for fungal prophylaxis, antimicrobial prophylaxis, avoidance of live vaccines for these patients as well as family genetic counselling.