THE BENEFIT OF EARLY DETECTION OF SICKLE CELL DISEASE IN IMPROVING PATIENT HEALTH PROGNOSIS AND REDUCING MORTALITY RATE IN NIGERIA

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Relevance. Sickle cell disease (SCD) is a genetic disorder which occurs as a result of mutated HbS. The resulting consequences of this disease are varying signs, symptoms and complications ranging from anemia, severe pain (crises), dactylitis/arthritis, eye damage, leg ulcers, aseptic necrosis/bone infarcts to fatal complications such as stroke and ultimately death. The main mechanism leading to hemolytic anemia and vaso-occlusive events is due to the replacement of glutamic acid by valine at the 6th position in the beta globin polypetide chain. This gives the affected red blood cells a characteristic sickle shape. The world atlas records Nigeria as the country with the highest disease incidence rate of about 91,011 born with SCD per year. This rate accounts for almost 2% of all new births annually. Most of these children are only known to have this disease in most cases only after emergence of complications and even death due to late diagnosis [2]. The estimated mortality rate is around 50-80% of children under 5 years of age. In countries like the United States of America where newborn screening and comprehensive care plans have been put in place, the mortality rate has been significantly reduced with survival rates of up to 18 years being 94% [1,3]. The World Health Organisation has estimated that up to two-thirds of the deaths caused by sickle cell disease are preventable by easy, inexpensive actions such as early detection after birth and early interventions through immunization against infections and timely response to acute events.

Aim of study. To estimate the rate of newborns affected with SCD in order to provide early necessary and timely interventions to improve life expectancy and reduce morbidity.

Methods. We made an analysis of the research that was conducted at Chukwuemeka Odumegwu Ojukwu University, Akwa, Nigeria from September 2013 to October 2017. The screening of SCD was done by determining the genotype through the use of isoelectric focusing eletrophoresis machine. In children who were found to have HbF and HbS, they were sent to pediatric sickle cell unit for follow up and management. The total number of new births within the four years amounted to 4961 of which 2410 were males and 2551 were females. Ages ranged from 0-30 days.

Results and discussion. Of the 4961 newborn children within the age range of 0-30 days of which 2410 were boys and 2551 were girls, 3733 (75.2%) had the hemoglobin AA genotype (1310 males, 2423 females), 1208 (24.3%) had the hemoglobin AS genotype (carriers) (583 males, 595 females), 16 (0.32%) of them had the hemoglobin SS genotype (7 males, 9 females) and 4 (0.08%) had hemoglobin AC genotype (1 male, 3 females) which is a variant of the AA genotype; having lysine substitute in place of glutamic acid.

Conclusions. Based on this study, it can be put forth that the availability of newborn screening for sickle cell disease for hospitals in Nigeria will help a great deal in proving early diagnosis as well proper follow up, prevention of exacerbating factors and management of associated events.

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MRI DIAGNOSIS OF REACTIVE ARTHRITIS

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Relevance. Diagnosis of reactive arthritis (ReA) today remains a difficult task, especially in cases where it is not possible to detect a "trigger" infection, rheumatoid factor is not detected in the blood, and the use of standard radiography is not informative [1]. In this regard, it is important to study the informativeness of such a modern sensitive diagnostic method as magnetic resonance imaging (MRI) for assessing joint lesions in patients with ReA.

Aim of study. Study of MRI features and signs of joint lesions in reactive arthritis.

Methods. We examined 12 patients with ReA: 4 women (30%), 8 men (70%). The median age of patients with ReA was 34 years (31-42). The median duration of ReA was 1 year (0.3-3.25). Among patients with ReA, there were 6 (50%) patients with acute ReA and 6 (50%) patients with chronic ReA. The control group consisted of patients who had no history or clinical symptoms of joint disease at the time of examination. Among them, 15 men (50%) and 15 women (50%). The median age in KG was 38.5 years (33-48). All patients underwent MRI examination of the knee joints in the T2 STIR mode (with suppression of the adipose tissue signal).

Results and discussion. As a result of MRI of the knee joints in patients with reactive arthritis, the following signs of damage to the entesial structures were revealed: thickening (93%), loosening (89%), the appearance of areas of reduced and increased density (94%), micro-breaks (defects) of fibers (78%), the presence of local osteitis