Oncology-hematology 7 Newborn screening for sickle cell disease : 13-year experience at the CHR "La Citadelle", Liège.

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Introduction:

As recently demonstrated (1), sickle cell disease (SCD) is the most frequent genetic disease in Belgium. Its detection during the neonatal period significantly reduces the mortality during infancy and early childhood (2). In fact, it enables clinicians to establish a specific management such as intensified immunization, penicillin prophylaxis and parent education.

Today, a universal neonatal screening is implemented in the USA (3), in the UK (4,5) and in the Netherlands (6). A selective screening is implanted in France (7).

Despite the recommendations from the World Health Organization and clinical pertinence of these programmes, screening for SCD is not yet approved by Belgian authorities.

Aim:

The purpose of this study is to relate our 13-year experience with a universal neonatal screening programme at the CHR Citadelle, and moreover to report the clinical outcomes of the patients who were diagnosed for SCD.

Methods:

Since September 2002, newborns have been systematically screened for SCD at CHR Citadelle regardless of ethnic background.

Cord blood samples are collected and analysed by capillary electrophoresis. Abnormal results are confirmed by HPLC.

Pathological results are rapidly reported to the referent haematologist paediatrician who organises the clinical management.

A family study is advised in all cases.

For each patient, the following clinical data were collected: number of nights of hospitalizations (NOH) per year, number of vasoocclusive crisis (VOC) per year and number of transfusions required (TR) per year.

Results:

From September 2002 to November 2015, a total of 30.659 newborns were screened. 33 SCD were diagnosed : 1 newborn per 1000 including 26 homozygous for HbS or compound heterozygous for HbS and ⊠°-thalassaemia (FS) and 7 compound heterozygous for HbS and C (FSC).

In addition, screening techniques allowed us to identify 627 HbS carriers and 141 other clinically significant Hb variant carriers (96 for HbC, 25 for HbE, 15 for HbD-Punjab and 5 for HbO-Arab).

Clinical data were available for all patients except one (FSC) who was lost to follow-up. The mean age at the end of the study was 53,82 months. The mean number of VOC per year was 0,63 (0,78 for FS and 0,04 for FSC). The mean number of NOH per year was 6,61 (7,64 for FS and 2,79 for FSC). The mean number of TR per year was 0,75 (0,94 for FS and 0,03 for FSC).

None patient died during the study except one (2,9%).

Conclusion:

The validity of a newborn screening programme for SCD has been clearly shown at the CHR Citadelle : for 2500 neonates tested per year, we screened 1 SCD per 1000 and identified 20 HbS carriers per 1000, i.e. a prevalence comparable to what is observed in Brussels, London or in Île-de-France. The programme including screening and diagnosis (offered by our laboratory for 13 years), genetic counselling, follow-up and care of patients, has made it possible to ensure a fast multidisciplinary management before the child leaves the hospital.

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