823

EPIDEMIOLOGICAL CHARACTERISTICS OF DOWN SYNDROME IN THE REGION OF WESTERN HERZEGOVINA

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During the 15 years period (1994-2008), 46 children with Down syndrome (DS) (31 boys and 15 girls) have been registered at the Neonatology Department of the University Hospital Mostar (Bosnia and Herzegovina). The average prevelence of DS for the region of Western Herzegovina (WH) (217 944 inhabitants) is 1,5 per 1,000 newborns in that period.

DS has been proved by means citogenetic analyses in 38 children. In 36 there was a regular type of trisomy 21 (94.7%) and in 2 the translocation type of DS was found.

33 parental couples (71.7%) were over 35 years of age. Early amniocentesis due to the parent´s age was made in two cases only, with positive dignosis. Usually, parents are refusing prenatal diagnosis due to religous reasons.

All children with DS have had five or more easily recognized minor malformations present in variable combinations. 28/46 had major malformations and three children have had mild forms of conatal hypothyreosis. Seven children (15.2%) died in neonatal or early infant period due to sepsis and complex cardiovascular anomalies. Considering evidently present major malformations and accompaning complications, DS remains very important issue for routine pediatric clinical practice.

824

CONGENITAL HAND DEFORMITIES AND ASSOCIATED SYNDROMES

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Background: Congenital Hand Deformity is an upper limb abnormality which present at the time of birth. Only 5% of congenital hand deformities occur as part of a recognized syndrome.

Objective: To retrospectively review all congenital hand deformities treated in Children University Hospital from January 2003 to October 2009 and correlate with syndromes associated.

Methods: Getting the list of hand surgeries from theatre. Retrieve charts from Chart Library. Compiled and enter data in Microsoft Excel.

Results: 20% patients have an associated syndrome. 29.3% presented with polydactyly. 7 of them are associated with syndromes. 59 out of 317 has syndactyly and 15% are associated with Apert Syndrome. 70 out of 317 are associated with radial ray abnormality and 24%has a syndromic background. None of the radial ray abnormality are associated with fanconi anaemia. 45 out of 317 presented with multiple ulnar deformities and 3 are associated with syndromes. 38 out of 317 presented with symbrachydactyly and 21 % are associated with various syndrome. 10 out of 317 are associated with multiple hand deformity and mostly they are associated with amniotic band syndrome. There are 2 patients that present with macrodactyly, however they are no medical association noted.

Discussion: In conclusion, 19.9% of the patients has associated medical background. None of them has proven Fanconi Anaemia manifestation. Fanconi Anaemia is commonly associated with radial aplasia.

Hence in the future, we will retrospectively screen all patient that has radial ray abnormality and prospectively screen them for fanconi anemia screening test.

825

TRISOMY 13 WITH ANORECTAL MALFORMATION:AN ASSOCIATION OR AN INCIDENTAL FINDING

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This report describes an infant with trisomy 13 associated with imperforate anus. Clinical features of patau syndrome described in standard texts do not include imperforate anus or anorectal malformations (ARM). In our search of the English