

dissimilarity index which indicate the percentage of the peak area of the fragment (operational taxonomic unit: OTU) was used.

Results Two fecal samples of 1 CC infant and 14 fecal samples of 7 BA infants were obtained. Nine predominant OTUs were detected with Bsl I digestion. The microbiota consisted of microbial communities of Bifidobacterium, Lactobacillales, Bacteroides, Prevotella, Clostridium clusters IV, XI, and XVIII, and Clostridium subcluster XIVa. The Bifidobacterium, Bacteroides, and Clostridium clusters were detected predominantly in CC than BA group. Lactobacillales was most predominant group in BA feces.

Conclusions Bacterial DNA showed marked differences in the composition of fecal microbiota in CC and BA infants. Molecular analysis of colonic microbiota using 16S rRNA gene libraries and T-RFLP might be useful to differentiate CC from BA.

734 ASSESSMENT OF DNA DAMAGE USING COMET ASSAY AND DETECTION OF OXIDATIVE STRESS PARAMETERS IN DOWN SYNDROME

doi:10.1136/archdischild-2012-302724.0734

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Background Down syndrome is one of the commonest numerical chromosomal aberrations. Recent studies showed that oxidative stress is an important pathological factor in Down syndrome.

Objective To estimate the level of oxidative stress and DNA damage in Down syndrome patients.

Patients and methods: Fifteen Egyptian patients clinically diagnosed and cytogenetically proven to have Down syndrome. Fifteen Egyptian healthy controls were recruited from the outpatient clinic of Clinical Genetics Department, National Research Centre. Oxidative stress parameters including total antioxidant capacity (TAC), Superoxide dismutase (SOD) enzyme activity and Malondialdehyde (MDA) biomarkers were estimated. DNA damage was determined using the alkaline comet assay.

Results The MDA and SOD levels in Down syndrome patients were significantly higher than control group ($p=0.000$ & 0.01 , respectively). Total antioxidants levels were non-significantly higher than control group ($p=0.54$). Statistical analysis of DNA damage levels in DS patients compared to controls showed significant increased levels ($p=0.000$). There was a positive correlation of DNA damage levels with age in DS patients but not reaching a significant value ($p=0.536$). A non-significant positive correlation was detected between DNA damage levels and both MDA and TAC levels ($p=0.8$ & 0.37 , respectively). Also a non-significant negative correlation of DNA damage levels with SOD levels was noticed ($p=0.14$).

Conclusion Oxidative stress plays a major role in DS pathogenesis.

735 CONTRIBUTION OF ALKYLATING AGENTS IN THE CYTOGENETIC DIAGNOSIS OF FANCONI ANEMIA

doi:10.1136/archdischild-2012-302724.0735

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Background and Aims Fanconi Anemia (FA) is an autosomal recessive disease characterized by heterogenous phenotype which includes a bone marrow failure, diverse abnormalities and increased predisposition to develop leukemia. The cytogenetic diagnosis of FA cells to bifunctional alkylating agents, resulting in greatly increased chromosomal breakage and radial structures induced by cross-linking agents. To estimate the sensitivity and the specificity of the Mitomycin C (MMC) and the Diepoxybutan (DEB), two alkylating agents used in the diagnosis of the FA, we studied the chromosomal

instability on 22 patients using variable types and concentrations of these alkylating (25 and 40 ng/ml of MMC, 0.1µg/ml of DEB).

Methods Heparanized venous blood samples were collected and were processed for the cytogenetic methodology in this study. After culture, 100 of metaphases were analysed to evaluate the frequency of chromosomal aberrations.

Results The MMC test at 25ng/ml was High sensitive for FA. The DEB test showed a better specificity. The study of the mitotic segregation of sexual chromosomes by FISH took away any abnormality of the segregation to cells FA.

Conclusions A molecular study of the sensitivity and the specificity of the alkylating agents used according to the group of complementation will come refine the diagnosis of FA by establishing a gold standard.

736 CLINICAL AND CYTOGENETIC STUDY IN 116 CHILDREN COHORT WITH SEXUAL AMBIGUITY

doi:10.1136/archdischild-2012-302724.0736

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Disorder of sex development may result from multiple etiologies, led to many physical impacts involving genital abnormalities. Differentiation of a testis or an ovary from a bipotential gonad is a complex developmental process where various genes and hormones interact. Thus, the causality of sexual ambiguity was supported by the presence of many chromosomal abnormalities, especially when sex determination genes were involved.

A careful clinical examination will primarily interest to define the sexual ambiguity Prader score and the whole genital and extra genital malformation context.

Our study focuses on cases referred to the department of Cytogenetics and Reproductive Biology CHU Farhat Hached for ambiguous genitalia cytogenetic exploration from January 2010 to February 2012.

The 116 patients studied with ambiguous genitalia of varying severity, 80% were younger than 10 years. 7.5% of them showed chromosomal abnormalities.

In 3 patients, the chromosomal abnormality involved the autosomes 4, 5 and 1 with a 4p deletion in Wolf-Hirschhorn syndrome, deletion 5p in Cri-du-chat syndrome and 1q deletion revealed by Comparative Genomic Hybridization-array. These anomalies are associated with loss of genital differentiation protein function. In 4 other patients abnormal sexual chromosome number was identified. For 86 children with normal karyotypes 46, XY, SRY gene molecular cytogenetic screening was performed. A deletion may in these cases explain the phenotype.

Identification of gene rearrangements causing urogenital birth defects can have an impact on the diagnosis and genetic counseling. Thus more study is necessary to find new candidate genes in signaling pathways regulating sexual differentiation.

737 MOLECULAR CYTOGENETIC AND COMPARATIVE GENOMIC HYBRIDIZATION ANALYSIS IN NEWBORNS WITH CONGENITAL HEART DISEASES

doi:10.1136/archdischild-2012-302724.0737

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Congenital heart diseases (CHD) are abnormalities that occur at birth and resulting from a developmental defect during cardiac