66

Syndrome of ocular, skeletal & abdominal abnormalities. R. M'rad, F. Mazzoul, N. Belguith, L. Ben Jemaa, N. Smaoui, H. Chaabouni. Service des maladies congenitales et hereditaires EPS Charles Nicolle Tunis Tunisia.

We describe two sisters who had a unique association of facial, ocular & skeletal defects and abdominal muscle hypoplasia. The two affected sisters are the fourth & the fifth sibs, born to healthy first cousin parents originating from Tunisia, indicating autosomal recessive inheritance. The family history is unremarkable. Many of these features overlap those previously found in other malformation syndromes. However, the constellation of defects observed in these patients appears to represent a OSA syndrome previously reported in 1996 by Mingarelli et al. In the best of our knowledge, it's the second report of this syndrome.

67

Genetic study of two Tunisian Ehlers Danlos syndrome type VI. R. M'rad, F. Maazoul, N. Belguith, L. Ben Jemaa, N. Smaoui, H. Chaabouni. Service des maladies congenitales et hereditaires EPS Charles Nicolle Tunis, Tunisia.

Ehlers Danlos syndromes (EDS) represent a clinically and genetically heterogeneous category of connective tissue disorders characterized by hyperextensibility of the skin and hypermobility of the joints.
Clinical genetic and biochemical grounds distinguish eight types. The type VI is characterized by specific biochemical defect of lysyl hydroxylase.
We report 2 cases of EDS type VI, clinically and biochemically confirmed. We focused our interest on the range of clinical severity in these patients and the comparison between our patient's clinical data and clinical findings reviewed in the literature.

68

Withdrawn

69

Analysis of 70 adult patients referred for genetics evaluation. S. Novak[1], M.S. Williams2, P.J. Levonian2, K.D. Josephson3, J.L. Williams3. 1Gundersen Lutheran Medical Foundation, 2Gundersen Lutheran Medical Center, 3La Crosse Regional Genetics Services Project, La Crosse, WI.

We present a series of adult patients referred for diagnostic genetic services. Patients referred for prenatal diagnosis, adult-onset neurodegenerative disease or cancer susceptibility counseling were specifically excluded from analysis. A total of 70 patients were identified. Information was obtained regarding age, gender, referral source, previous genetics evaluation, referring diagnosis, final diagnosis, diagnostic tests and recommendations. The patients were able to be grouped into 8 general categories: Multiple congenital anomalies (MCA)(13 pts.), Mental Retardation (MR)(12 pts.), Collagen-connective tissue disorders (12 pts.), Chromosomal abnormalities (9 pts.), Bone growth disorders (8 pts.), MR/MCA (6 pts.), Endocrine/metabolic (5 pts.) and miscellaneous (5 pts.). Patients and referring providers were not surveyed regarding utility of the evaluation. However, genetics notes were reviewed on all 70 patients and potential benefits of the evaluation were identified (examples include reassurance, anticipatory guidance, familial risk among others). All patients had at least one potential benefit, with the majority having several. This study represents one of the largest published groups of noninstitutionalized adult patients that have undergone comprehensive genetic evaluation. The role of the clinical geneticist as a member of the health care team in this population will be discussed.