P68 An evaluation of referrals for genetic investigation of short stature

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Short stature is a common condition in paediatric practice and yet the causes are heterogeneous. In Hong Kong, children with subnormal linear growth are screened out by primary health care system and then referred to hospital paediatric units where preliminary investigations would be carried out. Those apparently well but short children are also routinely referred to clinical genetic service where clinical evaluation and karyotyping are performed to exclude genetic causes.

Aim: To determine the proportion of genetic causes in referred cases of isolated short stature and assess the effectiveness of the current referring system for short stature.

Method: Retrospective: Both females and males, age between 0 to 16, who were referred because of isolated short stature were included. Case records were studied in respect to age, gender, referring diagnosis, medical history, birth size, family history including parental height, physical examination and investigation results especially bone age and karyotype. The growth parameters were plotted against the growth charts published by The Chinese University of Hong Kong in 1993. Cases with associated abnormalities like mental retardation and cases with chronic illness were excluded.

Results: 353 cases were studied and 135 had inadequate information for analysis. Among these referrals, hospital paediatric units and student health service accounted for 84%and 11% respectively and the rest came from child assessment centre, private doctors and maternal and child health centre. In the study group, 90% were girls and 80% of patients were shorter than third centile.

Among patients referred because of short stature, 32% had chromosomal abnormalities (majority were Turner syndrome), 5% had sydromal disorders like Nonnan syndrome and Russel Silver syndrome, 3% had skeletal dysplasia and 56% had constitutional short stature or physiological delay of bone age.

Conclusions:

1. Proper referral letters enclosing serial growth measurements and investigation results like endocrine work-up and bone age are essential in order to reduce unnecessary delay in establishment of diagnosis and wastage of resources in repeating investigations.
2. Screening of short stature by student health service is successful as they achieved a 24.3% pick up rate of chromosomal disorders, mainly Turner syndrome, among girls with isolated short stature.
3. Children with retarded growth velocity should be referred for genetic evaluation before their body height dropped to third centile as some patients with mosaic Turner syndrome, chromosomal abnormalities and Noonan syndrome are taller than the third centile.
4. In cases of familial short stature, pathological cause should be excluded before diagnosing constitutional short stature. For example, cases of hypochondroplasia and mosaic Turner syndrome affecting the mother and the child were documented in our study.

P69 Juvenile chronic arthritis in Chinese children: a review of the local experience

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Objectives: To review the clinical features of different subgroups of juvenile chronic arthritis (JCA) in Chinese Children.

Methods: All Chinese children (<15 years) with juvenile chronic arthritis seen between 1988 and 1998 in the Paediatrics Immunology Clinic, Queen Mary Hospital were reviewed retrospectively.

Results: There were 22 Chinese children suffering from JCA in this 10 year period. The female to male ratio was 1:0.9. The median age of onset was 6 years 3 months old (range: 5 month old to 13 year 8 months old). According to the classification of juvenile arthritis from International League Against Rheumatism task force (1997), 50% patients suffered from rheumatoid-factor negative polyarthritis (n=11, M:F ratio=1:2.7); 13.6% persistent oligoarthritis (n=3, all females); 13.6% extended oligoarthritis (n=3, M:F ratio = 1:2); 13.6% enthesitis arthritis (n=3, all males); 9% systemic arthritis (n=2, all males). None of the patients suffered from rheumatoid-factor positive polyarthritis. The extra-articular clinical features in descending order of frequency were fever (41%), chronic uveitis (13.6%) and skin rash (13.6%). Rheumatoid factor was negative in all patients. The antinuclear factor (ANF) was positive in 9 patients (41%). There were 3 patients (13.6%) with chronic uveitis and all of them were ANF positive. Hence one-third of ANF positive patients had chronic uveitis. Non-steroidal anti-inflammatory agents were given to all patients whilst prednisolone and methotrexate to 45% and 36.4% patients respectively. Eight patients suffered from growth retardation and two patients suffered from severe functional impairment.

Discussion: JCA consists of different subtypes which are different with respect to clinical features and prognosis. Rheumatoid Factor-negative polyarticular JCA seems to account for a higher percentage in Chinese (50%) than in Caucasians (30%). Methotrexate is the choice of drug as the second line therapy. Regular eye screening for uveitis is important, especially for ANF positive patients.

P70 Changing trends in the management of children with cleft lip and

P71 Fluorescence in situ hybridization in the investigation of 23 cases