SUMMARY

SUMMARY

While the advent of cardiac catheterization has revolutionized premortem cardiac diagnosis, it has not made a careful clinical evaluation obsolete. Rather the clinical examination and invasive study are complementary.

The history can provide important information useful in assessing the severity of cardiac condition, in recognizing possible complications and in identifying aetiologic factors.

A reliable diagnosis can be made from physical findings alone more importantly clinical findings are the basis for selecting patients for invasive study.

Thoracic X-ray films remain the basis for evaluation of the pulmonary vascularity, cardiac and specific chamber size and cardiac contour.

The electrocardiogram yields information about abnormal haemodynamic loads placed upon cardiac chambers and is the principal tool in identifying and diagnosing cardiac arrhythmias.

Echocardiography and nuclear cardiology procedures are having an increasing and beneficial role in the diagnosis and management of many paediatric cardiac abnormalities. Through

these noninvasive diagnostic methods both functional and structural information can be obtained which was only obtained by contrast angiography and cardiac catheterization.

Despite the extensive development of the above mentioned non-invasive techniques, cardiac catheterization remains a key in the diagnostic process for neonates, infants and children because it provides anatomic and haemodynamic information, which other diagnostic methods cannot, but the attitude should be that cardiac catheterization confirms diagnosis and not that catheterization establishes it.

The main value of electrophysiological investigations has been in locating accurately the origin of any arrhythmia, Holter monitoring in contrast is primarily directed at treatment and takes into account the role of autonomic nervous system in spontaneous arrhythmias and their response to treatment. On practical backgrounds it is more important to have an effective treatment than a clear understanding of the arrhythmia, but some particularly resistant cases necessitate sophisticated investigation for the planning of definitive medical or surgical treatment.

Approximately one third of children with congenital heart disease will have an associated non cardiac malformation and about 10 percent will represent a malformation syndrome careful

history taking and physical examination by the clinician is the most important step in the diagnosis. Recognition and understanding of such syndromes are essential for genetic counselling and proper patient management.

Autosomal imbalance in general, has a high incidence of cardiac anomalies, whereas sex chromosome imbalance except for Turner syndrome has a low incidence of heart defects.

Any patient with congenital heart disease with multiple anomalies, especially if there is low birth weight, a central nervous system abnormality, or a short fifth finger should have a karyotyping as a part of overall clinical evaluation.

Because of the high association of congenital heart disease, the clinician must have a high index of suspicion and even when no murmur is apparent, an electrocardiogram and chest roentgenogram should be obtained routinely in patients with Down syndrome. Delay in diagnosing congenital heart disease in these patients carries the risk of finding them inoperable when first diagnosed; development of pulmonary hypertension being a rather early finding.

Turner syndrome should be considered in any girl with coarctation of the aorta and short stature, the regular measurement of blood pressure is very important in those patients.

So far, only a few drugs, viruses or other environmental agents have been proved to be teratogenic for man, but a larger number are suspected and further epidemiologic studies will likely uncover others.

To properly evaluate environmental influences, information should be obtained routinely on women during pregnancy. Here we need to persuade our obstetrical colleagues to help us by their routine recording of information such as family and past history and social status.

From the clinical point of view, syndromes with skeletal defects may present first to surgeons who may not be aware of the association of these conditions with congenital heart disease which affects their surgical management.

In the group of conditions with prominent skin lesions three points should be stressed. In infants or children with neurological symptoms (with or without evident calcification of the skull X-ray) the presence of arrhythmias should arouse the suspicion of the possibility of a cardiac tumour being present. In addition neurological symptoms associated with signs of right sided obstruction should also alert the one to the possibility of a cardiac tumour being the underlying cause, these tumours as pointed out are associated with Tuberous sclerosis. Patients with neurofibromatosis, if pulmonary stenosis

is present, the valve is very likely to be of dysplastic character. This is important from the surgical point of view as the approach in this case will be more radical.

This also applies to pulmonary stenosis associated with Noonan's syndrome, the most common of the group with characteristic facies. In this group, observation alone may allow the recognition of the disorder.

The question whether or not Kartagner, polysplenia and other syndromes of abnormal visceral situs share a common pathogenesis seem to merit further study. The differential diagnosis of such conditions is very important being of widely different prognoses.

Inspite of the different cardiac manifestations seen in various types of connective tissue disorders it is obvious that all of them share a more or less common pathology. While this is manifested clinically by aneurysmal dilatation of large arteries which usually lead to aortic regurgitation in cases of Marfan syndrome, and osteogenesis Imperfecta, the opposite is seen in cases of recessive forms of Cutis Laxa and Pseudoxanthoma Elasticum, where deposition of the abnormal material causes narrowing of the lumen with manifestations of stenosis or ischaemia.

In the heritable disorders of metabolism, emphasis is placed on mitral insufficiency and isolated silent cardiomegaly. If either one of these two conditions is found in an infant or child, and the aetiology is not clear, a possible underlying inherited disorder of metabolism should be excluded. If present the basic metabolic disorder will usually determine the long term outlook of the case and certainly should modify the approach to the patient which should be conservative in such cases.

Sometimes, aortic regurge, involvement of coronary and peripheral arteries are presenting cardiovascular manifestations. However, they usually manifest in early adulthood.

In the group of neuromuscular disorders, three points should be emphasized. First, in Friedreich's ataxia it was believed that cardiac involvement was very common. Recently, it has been suggested that cardiac manifestations are essential before the diagnosis can be established. Secondly, the question arises whether cardiac myotonia actually exists or not. Thirdly, in certain cases with muscular dystrophy the cardiac involvement may be the first manifestation of the disease and only subsequently the neurological findings become evident. The latter may initially be very subtle and a high index of suspicion is necessary to make the diagnosis. Therefore, in cases with findings such as mitral valve prolapse, arrhythmias or conduction

disturbances in which subtle neurological features such as mild muscle weakness appear, the possibility of a muscular dystrophy should be considered and the patient investigated with this diagnosis in mind.

Although arrhythmic disorders are uncommon, the Paediatrician should consider the possibility of a cardiac origin for an unexplained seizure or syncope in a young child and part of the work up should include an electrocardiogram.