DWARFISM

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While there can be no scientific explanation for either Gulliver's Lilliputians or for the "Little folk" so beloved by our Irish friends, the following is a brief account of the causes of stunted growth. As the subject is so extensive the account itself must be "dwarfed".

Dwarfism is defined as a stunting of skeletal growth and as such does not include failure of sexual development which is termed infantilism. However in clinical practice it is unusual to have to distinguish between the two as they frequently occur together.

One should have no difficulty in recognising the obvious dwarf such as an achondroplastic or a cretin but as the majority present as a borderline failure to grow, it is important to be aware of the less obvious causes.

The diagnosis must be made without undue delay so that any treatment needed can be started before the epiphyses close, after which no further gain in height is possible.

The commonest causes for being below average size are racial and constitutional, thus the Mongol races tend to be smaller than the Caucasoids and small parents tend to have small offspring.

Any chronic illness in infancy and childhood will stunt growth and if the condition lasts up to the time of epiphyseal closure a small adult will result. Illnesses often responsible are bronchietasis, megacolon, bony tuberculosis, polycystic kidneys and mucoviscidosis. Congenital heart disease has a rather special place in that, tissue hypoxia leads to impaired growth. Malnutrition, while uncommon in civilised countries, is still a major problem for many of the world's population and is responsible for many stunted children. Lack of sufficient food, chronic infection and parasitism often work hand in hand. Kala azar, malaria, intestinal worms together with deficiency states such as kwashiokor are all responsible for stunted growth.

Specific causes for dwarfism may be loosely classified under endocrine; metabolic, bone diseases and hereditary conditions.

ENDOCRINE CONDITIONS

The Lorain-Levi or "Peter Pan" type of dwarf is probably due to a pre-pubertal shortage of anterior pituitary growth. They are normally proportioned but rarely over 48 inches tall. They are usually of a bright mentality but exhibit infantilism. There is no apparent shortage of corticotrophin or thyrotrophic hormone.

The cause of the pituitary failure is not often clear but some cases are due to non-functioning pituitary growths and others to supra-sellar tumours.

Cretinism and juvenile myxoedema produce persons of short stature who show in addition the other features of hypo-thyroidism — coarse skin, low mentality, generalised slowing of body activities and constipation. Cretins also have a large tongue, retroussé nose with sunken bridge and trident shaped heads. Unlike the other endocrine type of dwarfism cretinism is cureable provided and only provided the diagnosis is made within a few months of birth. There is no place for hormone treatment in older cretins as they are not improved men-

tally or physically and are often made more difficult to manage.

Stunting or early arrest of growth may occur in children showing precocious sexual development either due to adrenal cortical hyperplasia or suprarenal tumour or due to a virilising granulosa cell tumour of the ovary. The short stature is due to premature closure of the epiphyseal plates. Removal of the adrenal or ovarian tumour or supression of the hyperplastic adrenal cortex with cortisione *may,* if performed early enough, result in normal growth.

Frohlich's syndrome, which appears to be due to decreased production of gonadotrophins by a pituitary which is directly affected by a hypothalamic disorder, produces children showing sexual infantilism and marked mental torpor. There is a feminine distribution of fat and atrophy of the skin and hair. This syndrome may be caused by a craniopharyngioma or by a chromophobe adenoma of the pituitary but in most cases the cause is not found. True Frohlich's syndrome is a rarity but the name is erroneously applied to many obese children who are simply slow to develop sexually.

A variation of this last syndrome is the extremely rare condition described by Laurence, Moon and Biedl which in addition to the features of Frohlich's shows polydactly, retinitis pigmentosa and marked mental deficiency. This condition has a marked familial tendency.

Progeria, another condition of extreme rarity and unknown aetiology, is believed to be due to pituitary dysfunction. These patients show grotesque wrinkling and ageing of the skin in addition to the infantile and stunted appearance.

METABOLIC CONDITIONS

Rickets due to a shortage of dietary calcium and vitamin D is uncommon now but was a potent cause of dwarfism. Children seen now with the features of rickets are usually suffering from one of the secondary forms.

Renal rickets, which is better called renal osteodystrophy is really a conglomerate collection of renal diseases which cause alterations in calcium and phosporus metabolism. Almost any renal disease can cause sufficient damage to drastically alter the metabolism of these elements in a growing child. Blockage anywhere in the lower urinary tract such as neuromuscular dysfunction of the bladder neck which may lead to bilateral hydronephrosis; chronic pyelonephritis and nephrocalcinosis may all be responsible. Glomerular rickets occurs when the glomerular damage is sufficient to impair the excretion of phosphorus. Damage to the tubules or congenital defects of tubular function leads to a variety of conditions, the best known being Fanconi's syndrome in which there is an inability to reabsorb phosphorus. This leads to a low serum phosphorus and the excretion of calcium via the gut. The net result is a shortage of both elements for bone production. Other varieties consist of an inability to reabsorb either glucose or aminoacids or bicarbonate ions so that in addition to the bony abnormalities the patient may show glycosuria, aminoaciduria or a metabolic acidosis or all these. A combination of glomerular and tubular deficiency is present in Butler—Albright's syndrome which consists of dwarfing due to "renal rickets", hypochloraemic acidosis, precocious sexual development and a patchy pigmentation of the skin.

Conditions in which there is intestinal malabsorption, especially coeliac disease may give rise to secondary renal rickets as well as stunting growth directly.

A rare metabolic disease which causes dwarfism is the glycogen storage dis-

order of Von Gierke. Children suffering from any of the lipoid storage diseases are small but the other features such as mental deficiency, splenomegally and anaemia are of greater importance.

HEREDITARY AND CONGENITAL CONDITIONS

Mucoviscidosis or cystic fibrosis of the pancreas as it was called leads to a small stature as well as an increased liability to infection and to malabsorption.

A group of conditins arousing particular interest at present are those in which chromosomal abnormalities can be demonstrated. Mongolism or as it is better called Down's syndrome, is one such disorder and it has now been confirmed that these children have an extra chromosome. Cells from a normal human have 46 chromosomes arranged in pairs and each can now be recognised by special microscopic techniques but cells taken from hundreds of cases of Down's syndrome have been shown to contain an extra chromosome either as a separate unit or attached to a normal chromosome. There is reported a mongol child without an additional chromosome but the significance of this is not yet known. Despite this exception analysis of the chromosome number is likely to be a useful diagnostic tool in the future. Using similar methods the sex chromosomes can be identified and it is now possible to correlate the anomalous chromatin found in some patients with aberrations of their sex chromosomes. Turner's syndrome in which the person is stunted and shows webbing of the neck and is physically female can be shown to have the nuclear sexing of a male, that is XY.

BONE CONDITIONS

Achondroplasia is the commonest bone disease responsible for dwarfism and is incidentally responsible for the majority of dwarfs. The condition is strongly familial and primarily affects bone ossified in cartilage. The long bones fail to increase in length and the basal skull bones do not grow normally. The epiphyses are enlarged and the diaphyses abnormally short giving the unfortunate sufferer the characteristic short arms and legs with a normal trunk. Achondroplastics are of normal intelligence, are often genial and of normal fertility.

Fragilitas ossium is a condition of abnormal ossification, the child often showing multiple fractures at birth. The stunting of growth is mainly due to multiple fractures and telescoping of the long bones. These children often show blue sclerotics and their parents may have otosclerosis.

Two other osteodystrophies are also uncommonly responsible for dwarfism -- diaphysial aclasia and Morquio's disease, both are exceedingly rare and are diagnosed by their radiological appearance.

To complete the list of causes mention may be made of lipochondrodystrophy or Hunter's syndrome, more commonly called Gargoylism due to the characteristic ugly appearance of these children.

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