syndrome have not been reported previously; the low intracellular magnesium concentration found in this patient suggests that magnesium deficiency may be more common in Bartter's syndrome than indicated by serum magnesium levels alone.

Anorexia, apathy, and muscle weakness are common features of magnesium deficiency (Shils, 1969); all of these were present in our patient and improved after the introduction of dietary magnesium supplementation. The fever and excessive sweating also disappeared with magnesium supplementation. These are not recognized manifestations of magnesium deficiency; however, it is interesting to note that magnesium administration is effective in reducing the fever and sweating that occurs in patients with osteogenesis imperfecta (Solomons and Stynen, 1969). Before the magnesium therapy, his progressive clinical course was in keeping with the severe type of Bartter's syndrome; now, at 16 months of age, he is progressing normally apart from a diminished growth velocity. This course is most unusual for subjects with the severe infantile type of disease. It is concluded that intracellular depletion of magnesium may be responsible for at least some of the severe features of this disease which do not respond to more conventional therapy, and that measurement of muscle magnesium and/or a prolonged trial of magnesium supplementation should be undertaken in all subjects with the severe type of Bartter's syndrome.

Summary

Intracellular magnesium deficiency, with normal serum magnesium levels, was shown in an infant with the severe type of Bartter's syndrome. After the introduction of oral magnesium therapy, there was marked improvement in appetite and in muscle tone and strength; the intramuscular concentration of magnesium increased to a normal level. At 16 months he was progressing normally apart from a diminished growth velocity, and has had an unusually prolonged survival. It is concluded that intracellular magnesium depletion may contribute to the clinical manifestations and fatal course of this disease.

This study was supported by grants from the Human Growth Foundation and the General Clinical Research Centers Program of the Division of Research Resources, National Institutes of Health (RR-69). Also supported in part by USPHS Grant No. RO1-AM12432 from NIHAMD, and by Agricultural Research Service, U.S. Department of Agriculture, Grant No. 12-14-100-9941 (61).

REFERENCES


JOHN W. MACE,* K. MICHAEL HAMBIDGE,† RONALD W. GOTLIN, REUBEN S. DUBOIS, CLIVE S. SOLOMONS, and FRED H. KATZ

Department of Pediatrics, University of Colorado School of Medicine, and Veterans Administration Hospital, Denver, Colorado, U.S.A.

*Present address: Loma Linda University School of Medicine, Loma Linda, California, U.S.A.
†Correspondence to Dr. Kenneth Michael Hambidge, University of Colorado Medical Center, 4200 East Ninth Avenue, Denver, Colorado 80220, U.S.A.

Generalized hypermobility of joints

Arthrochalasis multiplex congenita

Generalized hypermobility of joints has been reviewed by Hass and Hass (1958) who described 5 patients in detail and called the condition arthrochalasis multiplex congenita; they noted that little attention had been paid previously to the orthopaedic manifestations and stressed that generalized hypermobility could exist without skin laxity; they suggested that it was an entity separable from Ehlers-Danlos syndrome and that joint involvement was very variable.

Carter and Sweetnam (1960) showed the strong
familial tendency to generalized joint hypermobility in relatives of patients presenting with habitual dislocation of joints. Carter and Wilkinson (1964), in a study of normal children, suggested criteria for establishing the diagnosis and showed a higher incidence in relatives of boys with congenital dislocation of the hip. Kirk, Ansell, and Bywaters (1967) took a different view, concluding that generalized joint hypermobility is simply an extreme variant of the normal. Penrose and Smith (1966) stated that children with Down’s syndrome often show a generalized hypermobility of joints.

We present another example of severe joint laxity associated with Down’s syndrome where arthrographic studies of both hip joints were carried out.

Case history

A boy was born by normal delivery at home on 11 May 1964. He was admitted to the Premature Baby Unit at this hospital because of low birthweight (2·1 kg). Features of Down’s syndrome were present, and the diagnosis was confirmed by chromosome analysis, which showed trisomy 21. His parents and 3 sibs were thought to be normal. There was no associated congenital heart disease and examination of his hips in the newborn period was normal. He was discharged at the age of 12 days. His development since has followed the usual course of Down’s syndrome, with delayed musculoskeletal and intellectual progress. (He sat at 18 months and walked at 32 months.)

Follow-up at yearly intervals was uneventful until October 1970, when his mother stated that for several months she had found him lying nearly every morning with his left hip acutely flexed and with a swelling apparent in the groin. By manipulating the thigh, she was able to correct the abnormal posture with an audible and palpable click; sometimes the manoeuvre was painful. Afterwards, he could run about normally. She thought his right knee also looked abnormal.

Examination at this time was difficult because of his lack of cooperation and no abnormality of the hip was found, but the right patella dislocated medially without discomfort. Other features included definite hyperextensibility of the metacarpophalangeal joints and thumbs, enabling the fingers to come parallel with the forearm and the thumbs to touch the wrists; there was 15° of hyperextension of the knees and both feet were hypermobile. There was muscle hypotonia and the skin was soft. Hernial orifices were normal, as were the tendon reflexes. His gait was plodding in character, but he could run and stand on either leg unaided.

In order to evaluate his mother’s history, he was admitted to hospital for observation. No incident took place as she described, and accordingly he was examined under a general anaesthetic. Then it was found that anteroinferior dislocation could easily be produced in both hips by thrusting downwards the femora, with the knees and hips flexed. Also, both patellae were unstable and, on the right side, medial dislocation and subsequent reduction occurred readily; on the left, lateral dislocation was incipient and could have been produced by only a little force.

Radiology. Hip arthrography was considered to be more informative than plain radiography. It would show the dynamics across the hip joints, the extent of dislocation, the direction and amount of force needed to produce such a dislocation, and, most important, the state of the capsule and other soft tissue components of the joint.

Consequently, arthrography of both hips was carried out via anterior approach under general anaesthesia, using the technique as described by Grech (1972a, b).

Both femoral heads were shown to be well developed and properly seated. The capacity of both capsules was found to be well above normal and up to 8 ml saline and contrast medium could be injected without an appreciable increase in the intracapsular pressure showing no back flow into the syringe on releasing the plunger. The anteroinferior dislocation could easily and repeatedly be shown in both sides by thrusting downwards the femora with the hips and knees flexed. The Fig. (a) shows both femoral heads out of acetabular sockets, but the labra do not appear to be inverted and no interposition of soft tissue is shown. Such a loose and wide displacement is due to marked elongation of the capsule and ligaments. Subsequent reduction was always easy and complete by relaxing the downwards push and extending the hips and knees into neutral position (Fig. (b)).

Fig.—(a) Bilateral hip arthrograms showing wide anteroinferior displacement of the femoral heads by thrusting on the femora with the hips flexed due to marked elongation of the joint capsule. (b) Bilateral hip arthrograms showing complete reduction of the dislocation with legs in neutral position.
Progress

No active measures were advised and the child was observed in the clinic. The episodes of dislocation described so clearly by his mother have decreased in frequency as far as the left hip is concerned and, during the last 4 months, only one episode has occurred. The cause of the improvement is not explained, other than supposing that a change in sleeping posture has been adopted by the child. The right patella dislocates regularly with every flexion of the knee, but no surgery is contemplated yet. The right hip and left knee remain lax but not unstable.

We have examined the sibs; the eldest boy (16 years) is normal; the second boy (14 years) and the third boy (7 years) have hyperextensible metacarpophalangeal joints, but all other joints are normal: their skins are soft in texture. The youngest child (a girl aged 3) and her parents are normal.

Comment

It is unlikely that this child has congenital dislocation of the hips, since no abnormality was detected in the neonatal period. Children with Down's syndrome often appear loose jointed because of the associated muscle hypotonia, but generalized joint hypermobility is also described in Down's syndrome (Penrose and Smith, 1966). Our patient fulfils the criteria suggested by Carter and Wilkinson (1964), and can be regarded as a mild case of arthrochalasis multiplex congenita, as described by Hass and Hass (1958). The gross joint laxity may, in this case, be the result of the combination of two predisposing conditions: the familial tendency as evidenced in his brothers, and the hypermobility associated with Down's syndrome. The late onset of recurrent dislocation of the hip and apparent spontaneous regression at the age of 8½ years, presumably due to changes in his sleeping posture, are also of interest. The arthrographic studies show the dynamics produced across the hip joints and the mechanics involved in producing the dislocation.

Summary

Clinical and familial details are given of a case of severe joint laxity where arthrographic studies of both hip joints were carried out to show the mechanics involved in producing the dislocation.

The various conditions presenting with generalized hypermobility of joints are briefly reviewed. It is concluded that this can be regarded as another case of ‘arthrochalasis multiplex congenita’.

Chronic lactic acidosis in association with myopathy

In 1962 Luft and his colleagues described a patient with a hypermetabolic state and a myopathy associated with partial uncoupling of oxidative phosphorylation within structurally abnormal muscle mitochondria. Other patients with myopathy and mitochondrial abnormalities have since been reported, but in only one was a raised plasma level of lactate noted (van Wijngaarden et al., 1967).

The following report describes a girl with a marked limb girdle myopathy, raised plasma levels of lactate, pyruvate, and alanine, and bizarre mitochondrial changes in the muscles.

Case report

The patient, a girl, is the second child of unrelated parents. Pregnancy, birth, and early motor development were normal. At 4½ years of age she developed increasing lassitude, weakness, and episodic vomiting.

On examination, aged 6 years, her height was 102 cm and weight 14 kg (both below the 3rd centile); there was marked weakness in the muscles of the trunk, shoulder, and pelvic girdles. She could not lift her arms above her head, could only climb upstairs on all fours, and could walk only 30 to 50 m without tiring. There was also slight weakness in the small muscles of the hands, and the muscles tired readily. No other neurological or other abnormalities could be detected. An edrophonium test for myasthenia was negative.

Plasma creatine phosphokinase level was 9.6 μmol/ml

REFERENCES


J. R. OWEN, R. A. ELSON, and P. GRECH* Northern General Hospital, Sheffield.

*Correspondence to Dr. P. Grech, Northern General Hospital, Sheffield S5 7AU.
Generalized hypermobility of joints: arthrochalasis multiplex congenita.

J R Owen, R A Elson and P Grech

Arch Dis Child 1973 48: 487-489
doi: 10.1136/adc.48.6.487

Updated information and services can be found at:
http://adc.bmj.com/content/48/6/487.citation

These include:

Email alerting service
Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

Notes

To request permissions go to:
http://group.bmj.com/group/rights-licensing/permissions

To order reprints go to:
http://journals.bmj.com/cgi/reprintform

To subscribe to BMJ go to:
http://group.bmj.com/subscribe/