TECHNICAL REPORT
25-60
業 績 報 告 書

PSEUDOHYPOPARATHYROIDISM AND HYPOTHYROIDISM: A CASE REPORT

HIROSHIMA

偽上皮小体機能減退症および甲状腺機能減退症の1症例

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ATOMIC BOMB CASUALTY COMMISSION
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TABLE OF CONTENTS
目　次

LIST OF FIGURES
挿入図一覧表

PSEUDOHYPOPARATHYROIDISM AND HYPOTHYROIDISM: A CASE REPORT

偽上皮小体機能減退症および甲状腺機能減退症の1症例

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INTRODUCTION

In 1939 Drake, Albright *et al*[1] set forth the criteria necessary for a diagnosis of chronic idiopathic hypoparathyroidism. These criteria include decreased serum calcium and increased serum inorganic phosphorus levels often associated with tetany, frequently with developmental abnormalities of the teeth, and with cataracts. In order to make this diagnosis there must be no evidence of osteomalacia, steatorrhea, or chronic renal insufficiency by which the altered serum calcium phosphorus ratio might otherwise be explained. It had been shown earlier[2] that prompt phosphorous diuresis followed the administration of parathyroid hormone demonstrating in normal subjects and in patients with hypoparathyroidism an ability to respond to the hormone. In 1942 Albright, Burnett *et al*[3] reported three patients who satisfied the above criteria except for the fact that the serum calcium level and urine phosphorus excretion were not altered by the intravenous administration of parathyroid gland extract. It was felt that these patients suffered from a different disease, designated pseudohypoparathyroidism, which was associated with a renal end-organ resistance to the action of parathyroid hormone rather than with the individual organism's inability to produce the hormone as in idiopathic hypoparathyroidism. Since that time, reports of 67 cases of pseudohypoparathyroidism have been published. Detailed observations on the cumulative findings of the patients thus far reported with pseudohypoparathyroidism have been presented in two recent reviews by Bronsky, Kushner *et al*[4] and Cohen and

緒言

1939年に，Drake, Albright 等[1]は慢性特発性上皮小体機能減退症の診断基準を発表した．この基準によれば，本症では血清カルシウムの減少および血清無機燐の増加があり，しばしばテタニーが起り，また歯の発育異常および白内障併発も多い．この診断を下すためには，血清カルシウム・燐比率の変化の原因となる骨軟化症，脂肪便または慢性腎臓機能不全の徴候があってはならない．古い報告によると[2]，健常者および上皮小体機能減退症の患者に，上皮小体ホルモンを投与すれば迅速な尿中燐排泄が起り，このホルモンに反応する能力があることが証明されている．1942年に，Albright, Burnett 等[3]が報告した3例では，上皮小体抽出物の静脈投与によって血清カルシウムおよび尿中燐排泄が変らなかった事を除いては，上述の基準に該当していた．これらの例症は，別の疾患であると考えられ，これを偽上皮小体機能減退症と名付けた．この疾患は，特発性上皮小体機能減退症におけるように，上皮小体ホルモンを生産することができないというよりはむしろ，上皮小体ホルモンの作用に対する腎臓終末器官の抵抗に関連していると考えられた．その後偽上皮小体機能減退症は67例報告されている．最近，Bronsky, Kushner 等[4]および Cohen Donnell[5]の2つの報告で今日まで偽上皮小体機能減退症について網羅された所見に関する詳細なる再検討が行なわれた．後者の報告では，10才の

Donnell.[5] In the latter paper the authors also reported the first authenticated occurrence of hypothyroidism and pseudohypoparathyroidism in a ten year old girl. In 1958 a case was reported[6] as the first instance of pseudohypoparathyroidism observed in Japan. This patient was an adult who had seizures and mental retardation, but who had a normal serum calcium and phosphorus. However, the report of this patient's failure to respond to parathyroid gland extract was questionable and it is difficult to be certain of the diagnosis.

It is the purpose of this report to present what possibly is the second case of pseudohypoparathyroidism known in Japan and to describe another documented coincidence of pseudohypoparathyroidism and hypothyroidism in a single individual.

HISTORY: A 13 year old boy was sent to the referral clinic of the Atomic Bomb Casualty Commission (ABCC) in Hiroshima for evaluation of mental and physical retardation and seizure episodes. The child, youngest of five children in the family, was born following a full term uncomplicated pregnancy. Details concerning early development were vague, but it was noted during infancy that he appeared smaller than normal, had an unusually large tongue, and was of a sluggish temperament. He sat unassisted, crawled, talked, and learned to walk later than his siblings had. Only eight or ten deciduous teeth erupted. Most of these were lost very early because of decay and breakage. Several permanent teeth appeared in each jaw but were only partially erupted and were characterized by the same fragility of their predecessors. The patient's skin had always been rough and dry; he had low tolerance to changes in environmental temperatures, and often complained of the cold in the warmest weather. Compared to other children he was very easily fatigued and did not enjoy physical activity or participation in games. He slept ten hours each night and seemed to require a morning and afternoon nap on most days. The child

女児において甲状腺機能減退および偽上皮小体機能減退症の合併が初めて認められた．1958年に日本における偽上皮小体機能減退症の最初の症例が報告された.[6] この患者は痙攣発作および精神発育遅滞の症状を有する成人であったが，血清カルシウムおよび燐は正常であった．しかし，この患者が上皮小体抽出物に反応を示さなかった点に疑問があるので，その診断は信頼し難い．

本報告の目的は，日本における第２番目の症例と思われる偽上皮小体機能減退症例を紹介し，なお偽上皮小体機能減退と甲状腺機能減退の合併が立証された記録を報告することである．

現病歴　13才の男児が，精神的・肉体的発育遅延および痙攣発作があるので診断および検査のため，広島原爆傷害調査委員会（ＡＢＣＣ）へ紹介された．本患者は５人兄弟の末っ子で，母親の妊娠中に特記事項はなく満期安産で出生した．初期発育に関する詳細ははっきりしていないが，幼児期に正常よりは小さいように思われ，舌が異常に大きく，感情鈍麻があった．坐るのも，這うのも，話すのも，歩くのも同胞よりは遅かった．乳歯は８ないし10本生えたのみでその大部分は，虫歯や欠損のため極く初期に脱落した．上顎と下顎にそれぞれ少数の永久歯が生えたが，その萌出状態は不完全で乳歯と同じように脆弱であった．患者の皮膚は生来荒れて乾燥し，環境の温度の変化に対する耐性は低く，暑い季節においてもしばしば寒いと訴えた．他の子供に比べて，患者は極めて疲労し易く，肉体的活動や遊戯への参加を好まなかった．患者は毎晩10時間眠り，通常午前および午後に睡眠を必要とするようであった．６才の時就学し，著しい精神的障害があったことが両親にも教師にもはっきりしていたけれども，心理的理由か

began school at the age of six. Although it was obvious to parents and teachers that he was markedly mentally handicapped, he was, for psychological reasons, kept with his class throughout primary and junior high school. By the age of 13 he had reached the 7th grade but was able to read only with the proficiency expected of a 3rd grader and was unable to calculate. When he was ten years old, the patient began having "seizures" described as a stiffening of the legs without loss of consciousness, causing him to fall while walking or running. These episodes lasted 20 to 30 seconds. Their frequency appeared to vary with the amount of physical activity. There were often 10 to 20 such attacks each day. Medical consultation was first sought at the onset of these seizures, three years before the patient was seen at ABCC. The diagnosis of hypothyroidism was made and an unknown amount of thyroid extract was administered for about a year with no change noted in the quality or frequency of the seizures. The patient did appear to be more alert and active during this period, however.

ら小学校および中学校を通じて，級友と一緒に進級した．13才の時には，中学一年になっていたが読書力は小学三年生程度に過ぎず，計算はできなかった．10才の時“痙攣発作”が起るようになり，意識喪失はないが歩行中または走っていると脚が硬直し倒れるという．これらの発作は20ないし30秒間続き，その頻度は肉体的活動の量によって変るようである．かかる発作が1日に10ないし20回起ることがしばしばあった．この患者がＡＢＣＣで受診する3年前にこれら発作が起った時に初めて医師の診察を求め，甲状腺機能減退の診断が下された．これに対して用量は不明であるが甲状腺剤が約1ヵ年投与された．しかし発作の性質または頻度には変化がなかったが，この間，患者は一層機敏で活動的であったように思われた．

FAMILY HISTORY: Both parents are alive and well, as are four older brothers and sisters. There is no known family history of congenital or hereditary disease, nor history of consanguinity. All other family members are of average intellect and physical stature.

家族歴　両親も4人の兄姉も健在である．先天性または遺伝性疾患の家族歴はなく，近親結婚もない．その他の家族は凡て普通の精神的および肉体的発育を示している．

PAST MEDICAL HISTORY: The birth weight was 2.951 kg. The patient had mild icterus neonatorum at the age of five days, severe gastroenteritis with dehydration at one year, and measles and mumps before the age of five. He always had been very susceptible to colds.

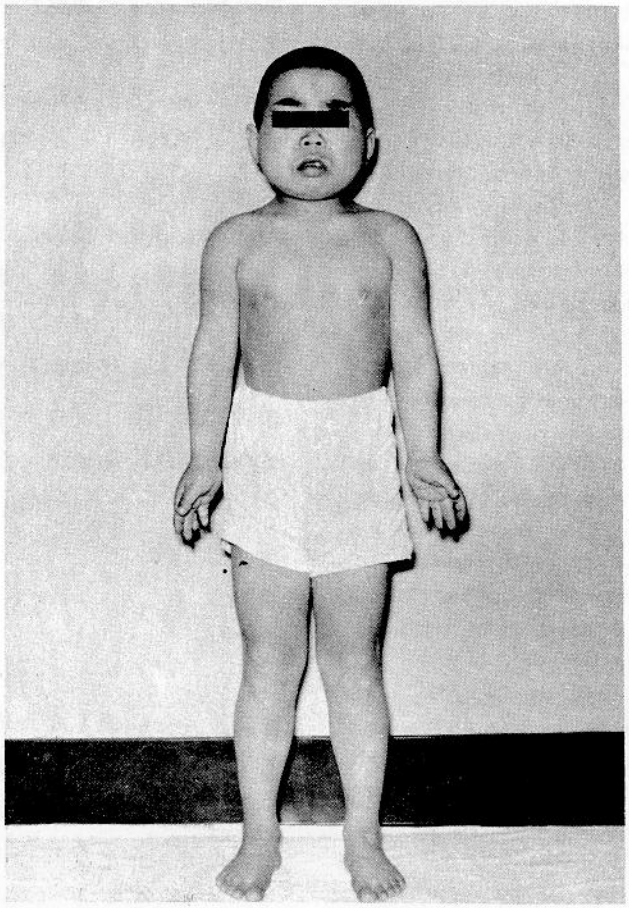
既往歴　生下時体重は2.95kgであった．生後5日目に軽度の新生児黄疸が現われ，1才の時に脱水症を伴なう強度の胃腸炎を発病した．5才の時までに麻疹および流行性耳下腺炎に罹患した．また常に風邪に罹り易かった．

PHYSICAL EXAMINATION: The child was a strikingly short, stocky boy (Fig. 1). He was dull-countenanced and slow-moving, but cheerful and cooperative. Both size and manner suggested a child two-thirds his age. The voice was deep and harsh. Height was 118.65 cm (average height for a 13 year old Japanese boy is 142.24 cm). Weight was

診察所見　身長の著しく低い，肥満した少年である（図1）．顔貌鈍，動作緩慢であるが，愉快で協調的である．体の大きさや態度から見れば，患者の年齢の3分の2位の子供に相当している．声は低く，しわがれている．身長 118.7cm（13才

FIGURE 1 SHORT, STOCKY BUILD, ROUND FACE, PROTUBERANT TONGUE

図1　身長は低く，肥満した体格，円い顔，突出した舌

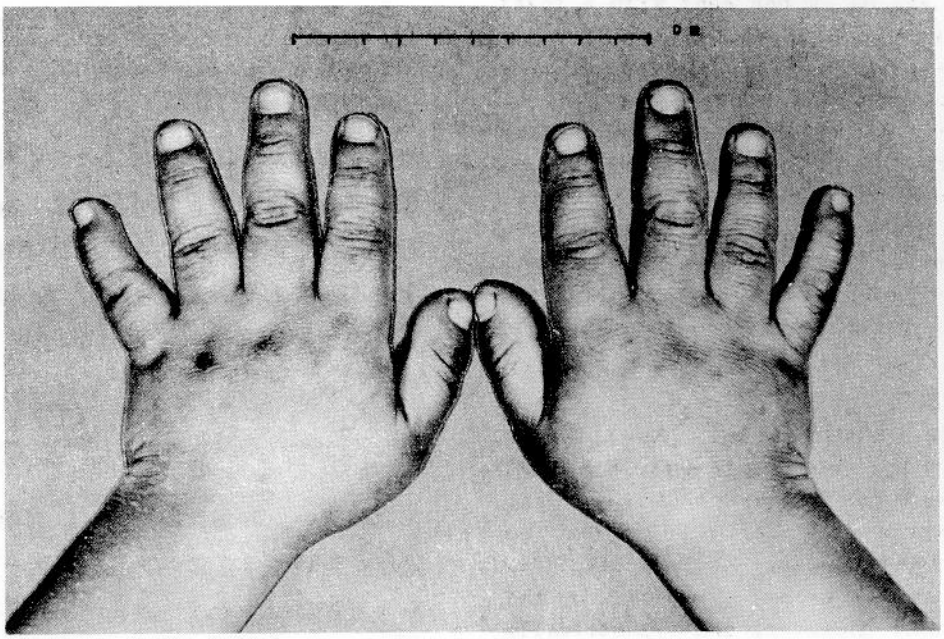


27.01 kg, blood pressure 90/65 mm Hg, temperature 32.2 C, and pulse rate 84. The skin was pale, dry, and seemed thickened with an unusual amount of subcutaneous tissue about the base of the neck, the axillae, and anterior chest. The face was round with a broad, flat nose and widely spaced eyes. The limbs appeared disproportionately short for the trunk; the hands and feet were broad with short, stubby fingers and toes (Fig. 2). The hair was coarse and eyebrows thin, especially in the outer quadrants. The pupils were round and regular, and there were no lenticular opacities. The optic discs were

の日本人少年の標準身長は142.2cm）体重27.01 kg，血圧水銀柱90／65mm，体温32.2°C，脈搏 84，皮膚は蒼白，乾燥し，肥厚しているようで，首の基底部の固り，腋窩および前胸部には皮下組織がよく発達している．顔面は円く，鼻の巾は広く低い．眼の間隔は広い．脚は軀幹の割には不均衡に短かく，手および足の巾は広く，指および趾は短かく太い（図２）．毛髪は硬く，眉毛は特に外側部において薄い．瞳孔は整円．水晶体混濁は認めない．乳頭の輪廓は両眼ともやや不鮮明であるが

FIGURE 2 BROAD HANDS, STUBBY FINGERS

図2　手は広く，指は太い



somewhat blurred bilaterally; otherwise the fundi were normal. The lips were thick and the tongue was enlarged and protuberant. Only a few carious, poorly developed and severely abraded teeth were present in both jaws. The thyroid gland was not palpable. The heart and abdomen were normal. The genitalia were typically prepubertal in development. Neurological examination revealed hypoactive deep tendon reflexes, and the Chvostek and Trousseau signs were positive.

その他の眼底所見は正常である．唇は厚く，舌は肥大，突出している．両顎に腐蝕した発育不良の歯を数本認め，強度に摩耗している．甲状腺は触れない．心臓および腹部は正常である．性器は定型的な思春期前の発育状態を示す．神経科的検査の結果，深部腱反射の減弱を認め，Chvostekおよび Trousseau 徴候を認める．

LABORATORY EXAMINATION: Complete blood count revealed a normochromic normocytic anemia with a hemoglobin of 10.6 gm per cent. The white blood count and differential were normal. The urinalysis was unremarkable with a negative Sulkowitch test. Stool examination and serological test for syphilis were negative. The serum calcium was 5.25 mg per cent and the serum phosphorus was 10.78 mg per cent. The serum protein bound iodine was 3.2 μg per cent. The thyroid radioactive iodine uptake was 4.4 per cent in 24 hours.

検査事項　血球計算の結果，常色素性正常赤血球性貧血を認め，血色素量は10.6g %，白血球数および白血球分類は正常．検尿の結果著変はなく，Sulkowitch 検査は陰性．検便および血清梅毒検査の結果は陰性．血清カルシウムは5.25mg%，血清燐は10.78mg%．血清蛋白結合沃度は3.2μg%．甲状腺の放射能性沃度摂取率は24時間で4.4 %．

The basal metabolic rate was -32 and -39 on two occasions. The total serum cholesterol was 143 mg per cent and the serum alkaline phosphatase 11.5 units. The serum bound iron was 34 μg per cent with a total iron binding capacity of 192 μg per cent. A Thorn test was negative. The urinary 17-ketosteroid excretion was 1.5 mg in 24 hours, and the urinary 17-hydroxy corticosteroid was 2.9 mg in 24 hours (the normal values for a child this age are 1.5 to 5.0 mg in 24 hours and 2.9 to 12.0 mg in 24 hours respectively). An FSH assay was positive for both 5 and 80 mouse units (the usual normal range is between 5 to 30 mouse units).

基礎代謝測定を2回行なって，それぞれ−32および−39であった．総血清コレステロールは143mg%，血清アルカリ性フオスフアターゼは11.5単位．血清結合鉄は 34μg %，　総鉄結合能は 192μg %．Thorn 検査は陰性．尿中17ケトステロイド排泄は24時間で 1.5mg，尿中17ヒドロキシ コルチコステロイドは24時間で 2.9mg（この年齢の子供の正常値はそれぞれ24時間で 1.5—5.0 mg および2.9—12.0mg）．濾胞刺激性ホルモン検査の結果，5 および80廿日ねずみ単位（普通の正常範囲は 5 —30 廿日ねずみ単位）．

RADIOLOGIC EXAMINATION: Radiologic findings of the chest were within normal limits except for a coarsened trabecular pattern in the scapulae. Films of the skull revealed a somewhat shallow pituitary fossa, which was normal, however in the anteroposterior diameter, and symmetrically thickened parietal bones were seen in the anteroposterior views. There were irregular calcific densities visible in the region of the basal ganglia. The extremities were short with short metacarpals and premature closure of the epiphyses. The lateral aspect of the shaft of the left radius was irregular and contained an ovoid area of cystic-appearing translucency 2 cm in length. Soft tissue calcifications were seen in the left upper and left lower extremities. Radiologic examination of the teeth revealed visible lamina dura but either severe enamel hypoplasia or defective dentine calcification was present in the canine and premolar teeth of both jaws. The deciduous incisors were present with no radiologic evidence of the germ of the permanent incisors or molars.

X線検査　胸部X線所見は，肩甲骨の粗造な梁柱像を除いては正常である．頭蓋X線撮影の結果，脳下垂体窩はやや浅いが腹背方向径は正常で，腹背方向撮影像では頭頂骨の左右対称的な肥厚を認める．脳底神経節部には不規則な石灰化陰影が見られる．四肢は短かく，中手骨も短かく骨端核の早期閉鎖を認める．左橈骨骨幹の外側面は不整形で，直径2cmの卵円形の嚢胞状透過性部分がある．左上肢および左下肢の軟部組織中に石灰沈着を認める．歯のX線撮影の結果，硬膜を認めるが両顎の犬歯および小臼歯には強度のエナメル形成不全または不完全な象牙質石灰化がある．X線上，乳切歯を認め，永久切歯または臼歯の歯胚の形跡はない．

ADDITIONAL EXAMINATIONS: The electrocardiogram was unremarkable except for prolongation of the S-T interval (often seen in hypocalcemia). The electroencephalogram revealed abnormally high voltage throughout, particularly marked over the occipital region, interpreted as a

その他の検査　心電図検査の結果S — T延長（低カルシウム血症によく見られる所見）を除いて著変を認めない．また脳波所見では，全般に異常な高電位差を認めるがこれは特に後頭部におい

diffuse nonspecific abnormality. The IQ (Weschler-Bellevue) was 49.

No increase in hourly urinary phosphorus excretion was noted (Fig. 3) in response to the Ellsworth-Howard test (200 units of parathyroid hormone administered intravenously preceded and followed by total hourly urinary phosphorus determinations).[2] Repeat thyroid radioactive iodine uptake was 3.6 per cent in 24 hours, after which 30 units of thyroid stimulating hormone (thyrotropar) was given intramuscularly over three days. On the fourth day, the thyroid uptake of radioactive iodine was 43.6 per cent.

CLINICAL COURSE (Fig. 4): The patient received daily administrations of 200,000 units of vitamin D, 6 gm of calcium gluconate and 1 gm of aluminum hydroxide gel (to delay the intestinal absorption of phosphorus). After 15 days, the serum calcium had risen to 6.5 mg per cent and the serum inorganic phosphorus had decreased to 9.5 mg per cent. The dosage levels were raised: 400,000 units of vitamin D; 9 gm calcium gluconate; and 2 gm aluminum hydroxide gel were administered daily for the subsequent two weeks. At this time the serum calcium had risen to a high level of 11.5 mg per cent and the serum phosphorus had fallen to 6.5 mg per cent. The repeat electrocardiogram revealed that the previously prolonged S-T segments had become normal. The high voltage waves did not appear in the repeat electroencephalogram and the tracings were interpreted as being within normal limits. There had been no further episodes of tetany; the Chvostek and Trousseau signs were not demonstrable. The child was more willing to take part in physical play and in general appeared to his parents to be more active and alert. Beginning 33 days following institution of therapy, the dosage of vitamin D was gradually decreased from 400,000 units daily to 50,000 units. Thyroid extract, one grain daily, was added to the regimen 71 days following the initiation of therapy. Three weeks after the addition of thyroid, the family noted

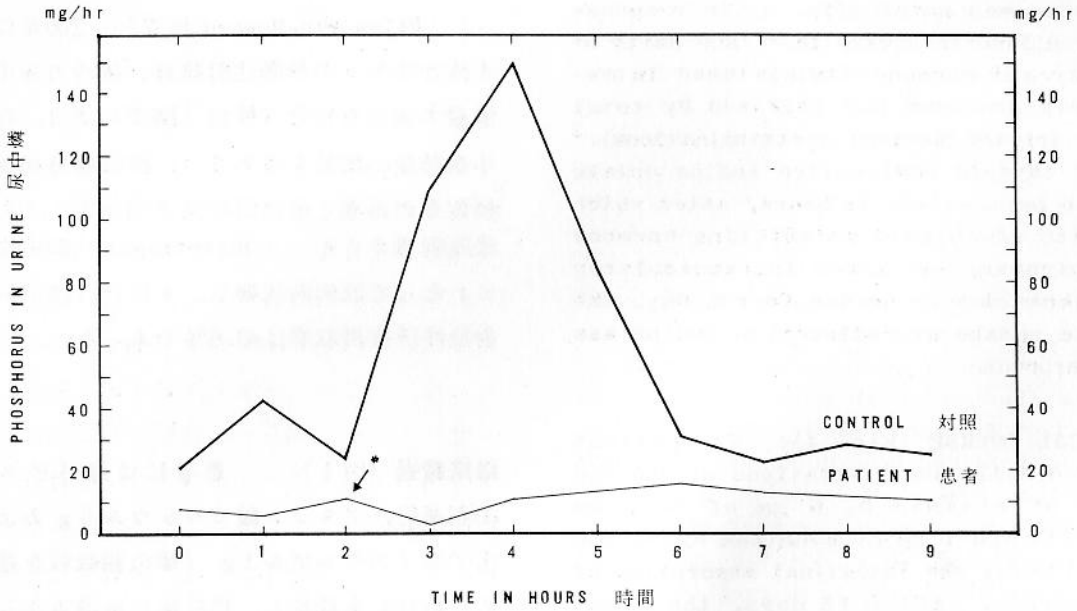
て著しく，瀰慢性の非特異的異常と解釈される．知能指数（Weschler-Bellevue）は49.

Ellsworth-Howard 検査[2]（200単位の上皮小体ホルモンの静脈注射前後，毎時の尿中燐総排泄量の測定を行なう検査（図３）では，毎時の尿中燐排泄の増加を認めない．甲状腺放射能性沃度摂取率の再測定値は24時間で 3.6%．その後，甲状腺刺激ホルモン（Thyrotropar）30単位を３日にわたって筋肉内注射し，４日目には甲状腺の放射能性沃度摂取率は43.6%であった．

臨床経過（図４）　患者には毎日ビタミンD 20万単位，グルコン酸カルシウム６g および水酸化アルミニウムゲル１g（燐の腸吸収を遅延させるために）を投与し，15日後に血清カルシウムは6.5mg%に上昇し，血清無機燐は 9.5mg%に減少した．投薬量を増量してその後２週間，毎日ビタミンD40万単位，グルコン酸カルシウム９g および水酸化アルミニウムゲル２g を投与したが，その結果血清カルシウムは11.5mg%まで上昇し，血清燐は 6.5mg%にさがった．そこで心電図再検査を行なった結果，以前に認められたS－T延長は正常になっていた．脳波の再検査では高電位差は消失し，正常であると解された．その後テタニーの発作はない．ChvostekおよびTrousseau徴候は証明されなかった．患者は肉体的な運動に参加することを一層好むようになり，全般的にみて一層活動的で機敏になったように両親には思われた．治療開始後33日目からビタミンDを１日量40万単位から５万単位まで次第に減量することにした．治療開始後71日目からは甲状腺剤を毎日6.5mg 投与した．甲状腺剤を追加投与してから３週間後に，家族のものは患者の運動に対する耐性が大いに増加したことを認めた．患者は日中に睡ることを止め，学校で活発になり注意を集中するようになっ

FIGURE 3 ELLSWORTH - HOWARD TEST

図3 ELLSWORTH - HOWARD検査

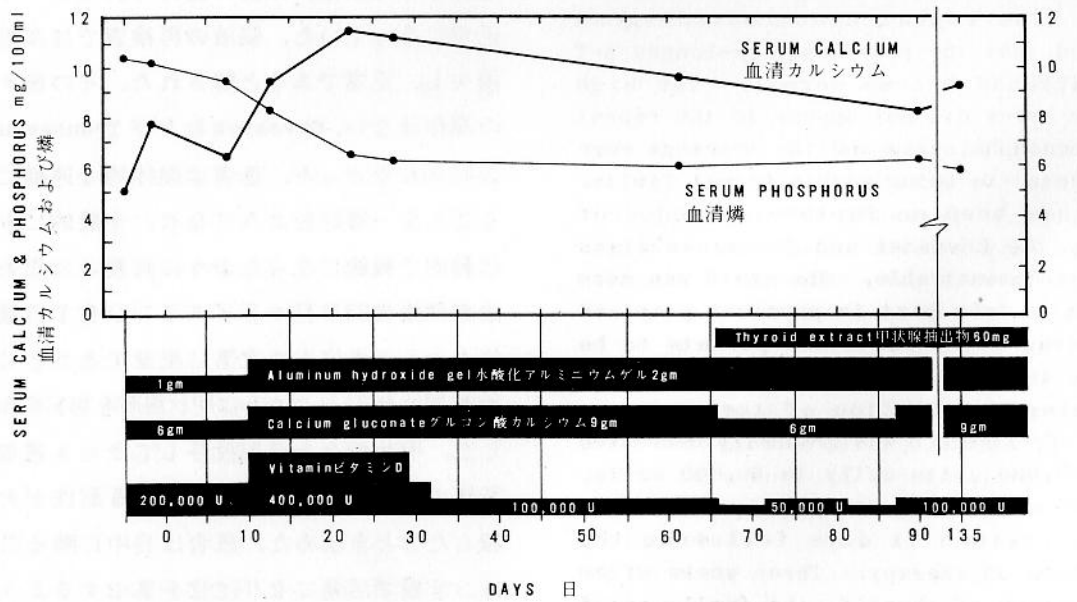


200 Units of parathormone administered intravenously.

200単位の上皮小体ホルモンの静脈注射

FIGURE 4 SERUM CALCIUM AND PHOSPHORUS IN RESPONSE TO TREATMENT

図4 治療に対する血清カルシウムおよび燐の変動



that there was a great increase in the patient's tolerance to activity. He had discontinued sleeping during the day and had become lively and attentive in school. He no longer complained of being cold in warm weather and for the first time had visibly perspired in response to the summer heat. At the time of this re-evaluation the patient certainly was more alert, but the appearance and physical examination were otherwise unchanged. The protein bound iodine was 6.4 μg per cent and the hemoglobin had risen to 12.0 gm. The serum calcium was 8.02 mg per cent and the phosphorus was 6.3 mg per cent. Only 50,000 units of vitamin D and 6 gm of calcium gluconate had been administered daily for the preceding three weeks. Although there had been no tetanic seizures during this time, nor were the Chvostek-Trousseau signs apparent, it was decided to raise the vitamin D to 100,000 units and the calcium gluconate to 9 gm daily. The thyroid extract was continued at the same dosage. The patient was seen again three months later (approximately seven months following initiation of therapy). He had been free of tetanic seizures and his general increased level of activity was considered quite satisfactory. There had been a 3.81 cm gain in height since his first visit. Physical examination revealed no other changes.

DISCUSSION

In addition to Drake's criteria for idiopathic hypoparathyroidism and failure to respond to parathyroid hormone, other findings have proved to be characteristic in patients with pseudohypoparathyroidism since Albright described the disease in 1942. Much of the present subject's history and findings are a summary of the characteristics most often observed. More than half of the patients reported have been of short, stocky habitus with round, moon-shaped faces. Brachydactylia with shortened metacarpals has been described many times, and more than two thirds of the patients with pseudohypoparathyroidism have

た．暖かい季節に寒いと訴えるようなことはなくなり，夏の暑い時には初めて眼に見えて汗をかくようになった．この時の診察において，患者は明らかに動作が機敏になっていたが，容貌や全身検査の結果それ以外の変化は認められなかった．蛋白結合沃度は6.4μg％で，血色素量は12.0g に上昇していた．血清カルシウムは8.02mg％，燐は 6.3 mg％であった．再診察の３週間前より，１日にビタミンＤ５万単位およびグルコン酸カルシウム６ｇを投与したのみであった．この間，テタニー発作はなく，Chvostek および Trousseau 徴候は現われなかったが，ビタミンＤを10万単位，グルコン酸カルシウムを９ｇ に増量することにした．甲状腺剤の用量はそのまま継続し，患者は３ヵ月後（治療開始約７ヵ月後）にもう１回診察を受けた．その間テタニー発作はなく，全般的に活動性の増加は極めて良好と考えられた．身長は初診時以来3.81cm 増加したが，その他の身体所見には変化はなかった．

考　按

1942年に Albright が偽上皮小体機能減退症を報告したが，その後本症には上皮小体ホルモンに対する反応の欠如と Drake の提唱した特発性上皮小体機能減退症の診断基準に合致する症状以外にその他の特徴的な所見が認められている．ここに報告した症例の病歴および所見の多くは，最もしばしば観察される特徴を網羅したものである．報告例の大半は，満月状顔貌を呈し身長の低い肥満した体質で，指は短縮し，中手骨も短い．偽上皮小体機能減退症例の３分の２以上にはある程

had some degree of mental retardation. As this patient was, most reported cases were initially investigated because of a chief complaint of tetany or convulsions. Other findings in many individuals with this disease and demonstrable in this patient are dyschondroplasia, premature epiphyseal closure, subcutaneous calcifications, dental defects with faulty enamel formation and failure of the adult teeth to erupt. The intracerebral calcifications in the region of the basal ganglia noted in this patient have been observed in about 40 per cent of those with pseudohypoparathyroidism. Specific objective neurological abnormalities are rare, but latent tetany (or a positive Chvostek sign) has been observed almost as often as hypocalcemia of which it is a reflection. Hyperphosphatemia, hypocalcemia in the absence of a phosphorus diuresis after intravenous administration of parathormone comprised the laboratory confirmation of the diagnosis of pseudohypoparathyroidism in this patient. He responded to the administration of vitamin D and calcium with a rapid reversion of the serum calcium-phosphorus ratio toward normal and concomitant complete amelioration of overt and latent tetany.

Hypothyroidism has been considered as a diagnostic possibility in many cases of pseudohypoparathyroidism, probably because of the similarity in appearance of patients with either of the two diseases. Shortness of stature, thickset habitus, moon face, delayed dental development, mental retardation, and sluggishness are common in children with either of these disorders. The simultaneous incidence of both diseases in one patient was not clearly demonstrated and reported until Cohen's and Donnell's case in 1960. In the patient investigated in the present report it was difficult to sort out all the findings characteristic of the two separate diseases, but this patient's intolerance to cold, easy fatigability, pale dry skin, infantile naso-orbital configuration, large tongue, and anemia are well known manifestations of childhood hypothyroidism. The depressed

度の精神発育遅延がみられている．大部分の報告例は，はじめテタニー又は痙攣を主訴としている．この患者にも認められたように本症を有する患者の多くは軟骨発育不全，早期骨端核閉鎖，皮下石灰沈着，エナメル形成不全を伴なう歯牙欠損および永久歯の欠如がある．この患者に認められた脳底神経節部分における脳内石灰化は，偽上皮小体機能減退症の約40%に認められている．特異的な他覚的神経科的異常はまれであるが，潜伏性テタニー（または Chvostek 徴候）は，その原因である低カルシウム血症と殆んど同頻度で認められている．この患者では高燐酸塩血症，すなわち上皮小体ホルモンの静脈注射後に尿中燐の排泄増加を伴なわない低カルシウム血症が認められたことによって，偽上皮小体機能減退症の診断が確認された．患者はビタミンDおよびカルシウムの投与に反応を示し，血清カルシウム・燐比は迅速に正常化し，それと共に顕在性および潜伏性テタニーは完全に回復した．

偽上皮小体機能減退症例の中で甲状腺機能減退症の疑いをもった例が多い．これは主としてこれらの患者の外観が酷似しているためであろうと思われ，その身長は低く，体質は肥満型で，顔貌は満月状を呈し，歯牙および精神の発育遅滞等が認められ，不活発であることが多い．この２つの疾患の合併例は，1960年に Cohen および Donnell の症例報告が行なわれるまでははっきりと証明されておらず，報告されていなかった．本患者においてこれら２つの異なる疾患のすべての特徴的所見をそれぞれ分類することは困難であったが，この患者の寒さに対する不耐性，疲れ易いこと，蒼白で乾燥した皮膚，乳児様顔貌，大きな舌および貧血は小児期甲状腺機能減退症のよく知られている現象である．臨床検査では基礎代射率の低下，

basal metabolic rate, low protein bound iodine, and radioactive iodine uptake provided convincing laboratory corroboration. A TSH test revealed findings similar to those observed by Cohen and Donnell. That is, there was an increase in the I^{131} uptake following stimulation with thyrotropic hormone, a ten-fold rise in this case, suggesting hypothyroidism secondary to pituitary failure to produce thyrotropic hormone. No other evidence of endocrine impairment was observed in this patient. A Thorn test was negative. Measurement of 17-ketosteroids and 17-hydroxysteroids showed low normal values for a prepubertal boy. Decreased levels of measurable corticosteroid hormones in hypothyroidism have been reported in the past.[7] The urinary FSH excretion was determined, revealing a positive test for 80 mouse units. The only physiological situations in which such a high value is normal are in females after the menopause and in normal preadolescent children at the time of the first spurt of gonadotropic hormone secretion.[8] The FSH levels are also elevated in primary hypogonadism,[9] which is of interest here because gonadal dysfunction, usually in the form of a delayed onset of puberty, is a common finding in childhood hypothyroidism[10] and has been recorded in several cases of pseudohypoparathyroidism.[11] Because of this boy's age, however, and the possibility that the elevated FSH level indicates approach of a normal pubescence further speculation regarding his sexual maturation must await the passage of time.

蛋白結合沃度の減少および放射能性沃度摂取率によって，ほぼ満足な確証が得られ，甲状腺刺激ホルモン検査の結果 Cohen および Donnell が観察したと同様の所見を認めた．すなわち甲状腺刺激ホルモン投与後 I^{131} 摂取の増加があり，この患者では10倍の上昇があったがこれは脳下垂体が甲状腺刺激ホルモンを生産することができないことに続発して起る甲状腺機能減退症を暗示するものである．この患者にはその他に内分泌障害の形跡は認めなかった．Thorn 検査を行なったがその結果は陰性であった．17ケトステロイドおよび17ヒドロキシステロイド測定の結果，思春期前の男児としては正常範囲内の低い値を示した．甲状腺機能減退症では測定可能のコルチコステロイドホルモンの減少があると以前に報告されている．[7] 尿中の卵胞刺激ホルモン排泄を測定したが80甘日ねずみ単位であった．かかる高い値が正常である唯一の生理学的状態は，閉経後の婦人および正常なる思春前期の子供において向生殖腺ホルモンが最初に生産される時にみられる．[8] 卵胞刺激ホルモン値は原発性性機能不全[9] でも上昇している．一般に思春期の遅延という形で現われる性腺機能障害は，小児期甲状腺機能減退症においてよく見られる所見であり，[10] 偽上皮小体機能減退症[11] の若干の症例にも報告されているので興味がある．しかしながら，この少年の年齢および卵胞刺激ホルモンの上昇が正常なる思春期の接近を示している可能性から考えて，本患児の性的成熟についての考察は時の経過を待たなければならない．

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