A Case of IgG Subclass Deficiency with the Initial Presentation of Transient Hypogammaimmuno-Globulinemia of Infancy and a Review of IgG Subclass Deficiencies

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Abstract

Primary immunodeficiency diseases are not common in children. The possibility of an immunological defect should be considered in any individual with repeated infections. A definite diagnosis for immodeficiency is sometimes difficult to achieve because of overlapping clinical manifestations. Immunoglobulin subclass deficiency is an immunological deficiency disease with which, one or more IgG subclasses are deficient. T cell immunity is normal. Patients may develop recurrent bacterial and respiratory infections or could remain asymptomatic[1].

Objective : The authors report a case of immunoglobulin G subclass deficiency presenting initially as transient hypogammaglobulinemia of infancy

Case report : A 2 month-old boy presented to Siriraj Hospital with a history of chronic protracted diarrhea, disseminated scabies and sepsis. On presentation, he had generalized scaly and maculopapular rash with no palpable lymph nodes. CBC revealed WBC 22,100 cells/cm^3 with PMN 42 per cent, lymphocytes 38 per cent, Eosinophils 4 per cent, Basophil 2 per cent and platelets 254,000/cm^3. The immunoglobulin levels were as follows: IgG 181 mg/dl, IgA < 6.6 mg/dl, IgM 26.3 mg/dl. Lymphocyte enumerations revealed CD_4 of 2,433 cells/cm^3 (N 1,460-5,160); CD_8 4,682 cells/cm^3 (N 650-2,450); CD_{19} 1,588 cell/cm^3 (N 500-1,500); CD_{16} 230 cell/cm^3 (N 573 $\pm$ 264). The initial diagnosis was X-linked agammaglobulinemia *vs* common variable immunodeficiency disease.

His diarrhea and five courses of sepsis responded well to antibiotics administration and courses of intravenous immunoglobulin (IVIG) replacement. His through IgG became normal at 2 years of age (after 12 months of IVIG). IVIG was stopped and the diagnosis was changed to transient hypogammaglobulinemia of infancy (THI). Nevertheless, during his 4 month follow-up he developed recurrent sinopulmonary infections (i.e, otitis media and pneumonia). Repeated immunoglobulin profile showed IgG 1,200 mg/dl, IgA 135 mg/dl, IgM 26 mg/dl, IgG subclass were IgG_1 1,030 mg/dl (N 280-830), IgG_2 30 mg/dl (N 40-2,400), IgG_3 22 mg/dl (N 6-130), IgG_4 3 mg/dl (N 3-120). A diagnosis of IgG_2 subclass deficiency presenting early as transient hypogammaglobulinemia of infancy was then made. Treatment with monthly IVIG was reinitiated and the patient is currently doing well.

Conclusion : The authors present a case of IgG subclass deficiency presenting as transient hypogammaglbulinemia of infancy. Follow-up of the immune profile and clinical manifestation is necessary for a definite diagnosis.

Key word : Immunoglobulin Subclass, Hypogammaglobulinemia

NETTAGUL R, VISITSUNTHORN N, VICHYANOND P
J Med Assoc Thai 2003; 86: 686-692

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Most immunodeficiency diseases have their distinct patterns of clinical presentations and laboratory findings. An early diagnosis of the immunodeficiency diseases in these patients is not easy as IgG subclass deficiencies may remain asymptomatic since several diseases such as X-linked agammaglobline-mia, transient hypogammagloblinemia of infancy have overlapping clinical manifestations. The pattern which is unusual for certain descant revise regurres requires reconsideration for another diagnosis.

IgG subclass deficiency is clinical situation in which patients have low levels (below 2 standard deviations) of age-adjusted geometric mean of one or more IgG subclasses(2).

Most frequently reported infections associated with IgG subclass deficiencies are recurrent upper respiratory tract infections, including sinusitis, otitis media and they usually do not develop life threatening infectious episodes.

A follow-up of clinical and laboratory investigations in patients is necessary for definite diagnosis.

CASE REPORT

Patient N.S. is a 2 month-old Thai boy referred to the Siriraj Hospital because of his chronic diarrhea. He was born by normal vaginal delivery with a birth weight of 2,500 g. He was breast fed for 1.5 months. His diarrhea developed after switching from breast to infant formula feeding. His diarrhea was resistant to medical treatments. Five days prior to admission, he developed a generalized pruritic maculopapular rash. On arrival at Siriraj Hospital, he was afebrile and was slightly pale. A scaly generalized maculopapular rash was noted. No tonsils or lymph nodes were noted on examination. Liver and spleen were not palpable. The laboratory work up was as follows Hct 25 per cent WBC 22,100 cells/cm^3 (N 42%, L 38%, E 4%, B 2%) plt 254,000 cells/cm^3. His stool contained numerous WBC but was negative for ova and blood sugar and electrolytes were normal. Scraping of skin lesions was positive for scabies. The anti-HIV (ELISA) was negative. TORCH titer was positive for anti HSV (IgG). His chest X-ray was normal.

Hospital course (Table 1)

During his 7 month's admission (from the age of 2 months to 9 months) five episodes of sepsis,

Table 1. Hospital course of this patient with multiple infections and the causative agents.

Age (month)	Diagnosis	H/C
2	Sepsis, Diarrhea	Candida, *S. aureus*
4	Sepsis, Diarrhea	Gram + ve cocci
6	Sepsis, O.M.	*S. aureus*
7	U.T.I.	*K. pneumoniae* (U/C)
8	Sepsis	*S. aureus*
9	Sepsis	No growth

Table 2. The immunoglobulin value on follow-up with clinical correlations.

Age (month)	IgG	IgA	IgM	Disease	H/C	IVIG (weekly)
2	400	40	30	Sepsis	NG	-
4	181	< 6.6	26.3	Sepsis	Gram + ve cocci	1
6	735	36	60	Sepsis + O.M.	S.aureus	1
7	700	20	30	U.T.I.	Klebsiella(urine)	2
8	695	14	48	Sepsis	S.aureus	3
9	640	28	-	Sepsis	-	2
12	630	28	86	-	-	6
19	930	78	51	-	-	6
24	1,280	30	125	-	-	Off
27	1,900	28	76	-	-	-

one episode of otitis media and urinary tract infection were observed. He was treated with several courses of systemic antibiotics. His diarrhea continued despite changes of formula from Alsoy to Olac and Pregestimil. Additional immunologic investigations were performed after 2 months of admission including normal PHA stimulation. There was delayed type hypersensitivity-no response to TB and tetanus. His repeated immunoglobulin showed IgG 181 mg/dl, IgA < 6.6 mg/dl, IgM 26.3 mg/dl. CD_4 was 2, 433 cells/cm^3, CD_8 4,682 cells/cm^3, B cell 1,588 cells/cm^3, The CD_4: CD_8 ratio was 0.5. His lymph node biopsy showed only a decrease in the number of B cells and plasma cells. Although this was not typical of XLA (presence of germinal center and plasma cell) the authors decided to initiate intravenous immunoglobulin administration at four months of age. His initial diagnosis was possibly X-linked agammaglobulinemia. (Table 2)

His immunoglobulin level gradually increased to near normal levels at two years of age also with improvement of clinical symptoms. His IVIG treatment was stopped at 27 months of age and the diagnosis was changed to transient hypogammmaglobulinemia of infancy (THI). During his 4 months follow-up (after discontinuation of IVIG), he developed recurrent sinopulmonary infections and pneumonia. Repeated immunoglobulin profile showed IgG 1,200 mg/dl, IgA 135 mg/dl, IgM 26 mg/dl, IgG subclass showed IgG_1 1,030 mg/dl (N 280-830), IgG_2 30 mg/dl (N 40-240), IgG_3 22 mg/dl (N 6-130), IgG_4 3 mg/dl (N 3-120). The diagnosis of IgG subclass deficiency was then made. Treatment with monthly intravenous immunoglobulin was reinitiated and the patient is currently doing well.

DISCUSSION

The patient presented with low levels of immunoglobulin G which became normal, at 19 months of age suggesting a diagnosis of Transient Hypogammaglobulinemia of Infancy (THI). Most patients with THI remain asymptommatic but could develop recurrent diseases such as otitis media, chronic diarrhea, bronchitis, urinary tract infection, osteomyelitis and cellulitis[3,4]. The presented patient could be an extremely severe case of THI due to his severe clinical courses during his early admission.

In general, patients with THI continue to have a low level of immunoglobulin after the physiologic hypogammaglobulinemia between 3 and 6 months of age. Immunoglobulin levels are normally achieved by the age of 18 to 36 months[2]. B cells are normal or near normal in number in such patients. The underlying pathogenesis for this disorder is unknown and could be heterogenous such as from deficiency of T helper cells[5]. Delayed functional maturation of B cells[6] has been postulated as contributing factors to the pathogenesis of THI. It is most commonly thought to be a variant of the normal, age-related acquisition of the capacity of B cells to produce immunoglobulins. Subsequent laboratory investigations should clarify such a problem. Kosnik reported a THI patient with streptococcal sepsis presenting as an acute abdomen[7]. The presented patient had 5 episodes of sepsis which is unusual for THI. Most THI patients do not require IVIG since infections are usually not serious[8]. The need for IVIG with frequent serious infections in the presented patient prompted the consideration for another diagnosis.

No THI patient who subsequently had a low level of IgG subclass after returning to normal level of immunoglobulin has been reported because it is a transient condition.

Specific antibody response to H. influenzae and Pneumococci vaccine should be determined in this patient since there is a general consensus that absence of one or more IgG subclasses is relevant only if this deficiency is associated with abnormal specific antibody response[9,10].

The mechanisms of IgG subclass deficiency are not clear. Deletions of sections of the immunoglobulin gene are extremely rare but have been found in a few healthy individuals with deficiencies of more than one IgG subclass in combination with IgA_1 deficiency[11]. The defect, therefore, probably resides at the level of the transcription of immunoglobulin gene in the B-lymphocyte. Abnormal T-lymphocyte control of B-lymphocyte function has also been proposed[12]. The mechanism which links THI to IgG subclass is unclear

IgG subclass is normally maintained within a relatively narrow range: IgG_1, 60-65 per cent; IgG_2, 20-25 per cent; IgG_3, 5-10 per cent; IgG_4, 3-6 per cent[13]. Each IgG subclass has an individual pattern of development with IgG_1 and IgG_3 attaining adult levels at an earlier age than IgG_2 and IgG_4[14]. IgG_3 deficiency occurs most often in adults, and IgG_2 deficiency is most common in children[15]. The clinical expressions and immunoglobulin levels in patients with deficiencies of one or several IgG subclasses are variable. Variability in the clinical expressions, types and degrees of deficiencies has led to the notion that this condition is a syndrome caused by more than one mechanism[9]. In general, patients with IgG subclass deficiencies usually do not present with life-threatening infections[2]. Asymptomatic individuals with IgG subclass deficiencies such as found in both blood donors[16] and among the relatives of patients with a variety of immunodeficiencies including family members of patients with IgG subclass deficiencies [17]. A wide range of infections with IgG subclass deficiencies have been reported such as pneumonia, bronchitis, otitis media, urinary tract infection, skin and eye infections. The predominant subclass deficiency report was IgG_1[18], whereas, others have generally seen deficiency of IgG_2[19] (and possibly IgG_4)[20]. The most common multiple immunoglobulin isotype deficiencies were combined IgG_2-IgG_4 [20] and combined IgG_2-IgG_4-IgA deficiency[18, 21]. Such combinations may be due to coordinate regulation of immunoglobulin isotype-switch expression since these heavy-chain loci are sequentially adjacent on chromosome 14[22,23]. Patients with recurrent and/or some respiratory infections with borderline IgG levels could have IgG subclass deficiencies. Patients with combined IgG-IgA deficiencies are predisposed to recurrent respiratory tract infections[18]. Isolated IgG_4 subclass deficiency with recurrent infections have also been reported[20]. Patients with IgG_1 and IgG_3 deficiencies may develop recurrent respiratory tract infections resulting in deafness (from chronic otitis media) and bronchiectasis particularly when associated with IgA deficiency[24]. Allergic patients may have an increased prevalence of low IgG values compared with non atopic individuals[25]. Low IgG_4 has frequently been observed in allergic patients resulting in recurrent infections more atopic control subjects with normal IgG subclass levels[20,26]. IgG subclass deficiencies, predominantly of IgG_2, has been observed in one third of children under 7 years of age presenting with severe asthma presumably due to delayed maturation of the immune system[27] or from chronic steroid usage[28-31]. The pattern of pneumococcal antibody response could be divided into three groups according to the postvaccination antibody titer, i.e. Gr sup I: Non responder, Gr sup II: Intermediate responder, and Gr sup III: Normal responder[32].

IgG subclass deficiencies have been observed in patients with vasculitides such as with Henoch-Schonlein purpura, autoimmune cytopenia, diabetes mellitus type[33,34], Freidreich's ataxia, Ataxia telangiectasia[35] and SLE[36] and patients with severe combined immunodeficiency, chronic granulomatous disease, chronic mucocutaneous candidiasis and deficiency of major histocompatibility antigens (Bare lymphocyte syndrome)[37,38] Initial treatment of patients with recurrent infections with low IgG subclasses and/or a selective antibody deficiency could be prophylactic antibiotics. The usual choices of antibiotics are amoxycillin and Trimetroprim-Sulfamethoxazole given twice daily. Failure to prophylactic treatment could justify the use of IVIG particulary those with specific antibody deficiencies. Duration of given IVIG is variable depending on the age and clinical course of individual patients[39].

Available IVIG preparations generally contain normal proportions of IgG subclasses but differ mainly in IgA contents. The recommended dosage is 200-500 mg/kg of IgG every 3-4 weeks. Dosages and

intervals can be adjusted according to severity of infections[40]. The mode of action can not be attributed to replacement of the respective isotypes of IgG, but is probably due to the effect of IVIG in preventing repeated viral infections. The biologic activity of the immunogloblin approved for treatment is more important than substitution of the respective isotypes because IgG subclass level is not significantly changed after the infusion[41].

Most patients with IgG subclass deficiencies have normal IgG levels due to a compensatory increase in other IgG subclasses. Total IgG concentrations could be low in IgG_1 deficiency due to a large contribution of this subclass[39]. The interpretation of low IgG subclass levels could sometimes be difficult due to a variation in IgG subclass concentrations with age and lack of universally standardized methods of assay. This pattern of seroconversion is characteristic of young infants aged 6 months or less[42].

The duration of IVIG treatment in the presented patient depended on his clinical course. The clinical course in the presented patient indicated that a definite diagnosis of his immunodeficiency could only be made by close follow-up overlapping the clinical manifestations.

(Received for publication on September 28, 2002)

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รายงานผู้ป่วยภูมิคุ้มกันผิดปกติชนิดพร่องไอจีจี ที่มีอาการแสดงของโรคช่วงแรกคล้ายโรคขาดภูมิคุ้มกันชั่วคราวในเด็กและทบทวนบทความ

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โรคภูมิคุ้มกันบกพร่องแต่กำเนิดพบได้น้อยในเด็ก ควรคิดถึงภาวะภูมิคุ้มกันผิดปกติในเด็กที่มีการติดเชื้อบ่อย ๆ การวินิจฉัยโรคบางครั้งยากเพราะอาการแสดงออกของโรคที่คล้ายกัน ภาวะภูมิคุ้มกันผิดปกติชนิด Immunoglobulin subclass deficiency (IgG subclass deficiency) เป็นภาวะที่ขาด IgG subclass ตัวใดตัวหนึ่งหรือหลายตัวโดยที่หน้าที่ของ T cell ปกติ ทำให้มีการติดเชื้อทางระบบทางเดินหายใจบ่อย ๆ หรือ อาจไม่แสดงอาการผิดปกติแต่อย่างใด

วัตถุประสงค์ : ได้รายงานผู้ป่วยภูมิคุ้มกันผิดปกติชนิด IgG subclass deficiency ที่อาการแสดงครั้งแรกคล้ายกับโรคขาดภูมิคุ้มกันชั่วคราวในเด็ก (Transient hypogammaglobulinemia of infancy)

รายงานผู้ป่วย : เด็กชายอายุ 2 เดือน มารักษาที่โรงพยาบาลศิริราชเรื่อง ถ่ายเหลวเรื้อรัง, เป็นหิดทั่วตัว และติดเชื้อในกระแสเลือด ตรวจร่างกายพบผื่นแดงทั่วตัวแต่ไม่พบต่อมน้ำเหลืองโต ผลเลือด CBC พบ WBC 22,100 cells/cm^3 PMN 42%, ลิมโฟไซต์ 38%, อีโอซิโนฟิล 4%, เบโซฟิล 2%, เกล็ดเลือด 254,000 เซลล์/ลบ.มม. ระดับอิมมูโนโกลบูลิน : IgG 181 mg/dl, IgA < 6.6 mg/dl, IgM 26.3 mg/dl CD_4 2,433 cells/cm^3 (ปกติ 1,460–5,160), CD_8 4,682 cells/cm^3 (ปกติ 650–2,450), CD_{19} 1,588 cells/cm^3 (ปกติ 500–1,500), CD_{16} 230 cells/cm^3 (ปกติ 573 $\pm$ 264) การวินิจฉัยเบื้องต้นคือ X-linked agammaglobulinemia หรืออาจเป็น common variable immunodeficiency disease

ผู้ป่วยเกิดอุจจาระร่วงและติดเชื้อถึง 5 ครั้งซึ่งตอบสนองดีต่อยาปฏิชีวนะและการให้อิมมูโนโกลบูลิน (IVIG) ระดับ IgG ของผู้ป่วยกลับมาเป็นปกติ เมื่ออายุ 2 ปี (12 เดือน หลังให้ IVIG) ซึ่งได้หยุดให้ IVIG และวินิจฉัยเป็น Transient hypogammaglobulinemia of infancy 4 เดือนหลังจากการติดตามพบว่าผู้ป่วยมี หูอักเสบและปอดบวมถึง 4 ครั้ง ตรวจระดับเลือดพบ IgG 1,200 mg/dl, IgA 135 mg/dl, IgM 26 mg/dl, IgG subclass พบ IgG_1 1,030 mg/dl (ปกติ 280–830), IgG_2 30 mg/dl (ปกติ 40–2,400), IgG_3 22 mg/dl (ปกติ 6–130), IgG_4 3 mg/dl (ปกติ 3–120) จากผลดังกล่าวทำให้วินิจฉัยผู้ป่วยรายนี้เป็น IgG_2 subclass deficiency ซึ่งอาการแสดงเริ่มแรกเหมือน Transient hypogammaglobulinemia of infancy อาการผู้ป่วยดีขึ้นหลังรักษาด้วย IVIG ทุกเดือน

สรุป : ได้นำเสนอผู้ป่วย IgG subclass deficiency ที่มาด้วยอาการคล้ายกับ Transient hypogammaglobulinemia of infancy การเฝ้าติดตามอาการและผลเลือดทางอิมมูน จะช่วยในการวินิจฉัยได้ถูกต้อง

คำสำคัญ : ภาวะพร่องไอจีจี, โรคขาดภูมิคุ้มกันชั่วคราวในเด็ก

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