Case Report / Olgu Sunumu

From The Perspective of The Core Competency of Community Orientation in Family Medicine: Two Cases of Gilbert Syndrome in The Same Family

Aile Hekimliğinde Toplum Odaklılık Temel Yetkinliği Perspektifinden: Aynı Ailede İki Gilbert Sendromu Vakası

Hacı Ahmet Aydemir

Dr. Filiz Dolunay Family Health Center, Erzurum, Türkiye

Abstract: Jaundice, which reflects increased levels of bilirubin in the blood, is one of the most important symptoms of liver disease and hemolytic disorders. Bilirubin levels can also be elevated in inherited disorders of bilirubin metabolism. Gilbert's syndrome is one of the most common of these inherited disorders. It is inherited as an autosomal dominant trait. It is also known as benign hyperbilirubinemia, in which indirect bilirubin shows a moderate course. These patients do not have associated liver disease. In this case report, two cases of Gilbert's syndrome in two siblings in one family, followed up in a family medicine unit, are presented and discussed from the perspective of community orientation, which is one of the core competencies of family medicine.
Keywords: Community orientation, Gilbert's syndrome, family practice, core competence

Özet: Kandaki bilirubin seviyesinin artışını yansıtan sarılık, karaciğer hastalıkları ve hemolitik bozuklukların en önemli belirtilerinden biridir. Bilirubin seviyeleri, bilirubin metabolizmasının kalıtsal bozukluklarında da yükselebilir. Gilbert sendromu bu kalıtsal bozuklukların en yaygın olanlarından biridir. Otozomal dominant bir kalıtım gösterir. İndirekt bilirubinin ılımlı bir seyir gösterdiği iyi huylu hiperbilirubinemi olarak da bilinir. Bu hastalarda eşlik eden karaciğer hastalığı yoktur. Bu olgu sunumunda, bir aile hekimliği biriminde takip edilen, bir ailede iki kardeşte Gilbert sendromu saptanan iki olgu sunulmuş ve aile hekimliğinin temel yetkinliklerinden biri olan toplum odaklılık perspektifinden tartışılmıştır.
Anahtar Kelimeler: Toplum odaklılık, Gilbert sendromu, aile hekimliği, temel yetkinlik

ORCID ID of the author: HAA. 0000-0001-9656-5089

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Correspondence: Hacı Ahmet AYDEMİR - Dr. Filiz Dolunay Family Health Center, Erzurum, Türkiye
e-mail: ahmetaydemir29@gmail.com

1. Introduction

Bilirubin is the end product of heme metabolism(1). Initially, elevated bilirubin was considered as a sign of liver disease. However, moderate elevation of bilirubin has been shown to be a protective mechanism in diseases caused by oxidative stress, particularly in atherosclerotic conditions(1). This protective function appears to be related to the protective effect of bilirubin on the vascular endothelium. High levels of bilirubin are known to reduce levels of advanced glycation end products, which are responsible for endothelial dysfunction(2).

Gilbert's syndrome is a benign condition of mild indirect bilirubin elevation in the absence of underlying hepatic and hemolytic disease, affecting approximately 10% of the world's population(3). It is caused by a congenital mutation in the enzyme uridine diphosphate (UDP) glucuronyl transferase, which is primarily involved in bilirubin metabolism(3). Due to the known antioxidant and hormone-like effects of bilirubin, people with Gilbert's syndrome have a natural resistance to cardiovascular disease, some cancers, autoimmune and neurodegenerative diseases. Diagnosis is made by excluding the presence of underlying liver disease or hemolysis. Evidence of a UDP-glucuronyl transferase polymorphism is not required for diagnosis, and the moderate elevation of bilirubin in Gilbert's syndrome does not require treatment(3).

Primary care is important because it is the patient's first point of contact(4). The financial burden of health care costs has increased the role of primary care in health care delivery, and primary care is central to the organization of health care services(5). It is known that in countries where primary care is strong, society's health problems are solved more quickly and easily, and inequalities in health care are kept to a lower level(6). With strong primary care, referral rates decrease and patients are more satisfied with the resolution of their health problems(7). In addition, hospitalization rates and all-cause mortality rates decrease because of the central position of the health system(8, 9). In view of this, it is important that physicians responsible for the management of primary care are trained to have the competence to undertake this important task. The implementation of the discipline of family medicine in a more qualified manner is achieved through the assimilation of the core competencies of this discipline and its implementation in the field under the guidance of these competencies. The discipline of family medicine has a total of six core competencies and twelve basic characteristics based on these competencies(10). Primary care management is one of these core competencies. This core competency places an important obligation on the family doctor to provide effective and appropriate health care.

2. Cases

Case 1

A 10-year-old girl was brought to our Family Health Center outpatient clinic by her mother with the complaint of periodic jaundice of the skin and sclera. She had no history of any disease. She was fully immunized and had no history of atopy. Arterial blood pressure: 90/60 mm/Hg, pulse: 82 beats/min, temperature: 36.2°C and oxygen saturation: 99%. Height: 143 cm (50-75 percentile), and weight: 26 kg (3-10 percentile). There was no hepatosplenomegaly on physical examination. Traube was open. Other systemic examinations were normal. The results of hemogram and biochemical tests are shown in Table 1.

At the examination of the patient approximately 2 weeks later, jaundice was not observed in the sclera and skin and the complaints at the first presentation had improved. No laboratory tests were performed at the next admission.

Case 2

An 18-year-old male patient presented with complaints of increasing weakness and fatigue over the past month and periodic jaundice of the skin and sclera. There was no history of any chronic diseases. There was no history of atopy in the patient whose immunizations were complete. Arterial blood pressure was 110/70 mm/hg, pulse rate was 71 beats/min, temperature was 36.6°C and oxygen

saturation was 98%. Height: 173 cm (25-50 percentile), weight: 54 kg (<3 percentile). There was no hepatosplenomegaly on physical examination. Traube was open. Other system examinations were normal. The blood test results were given in Table 2.

The family history of both patients showed that they belonged to the same family and that the father had migraine. There was nothing else unusual in the family history.

At the examination of the patient approximately 3 weeks later, jaundice was not seen in the sclera and skin and the complaints at the first presentation had almost completely resolved.. No laboratory tests were performed at the next admission.

Table 1. The results of the blood test analyses of Case 1.

Test Name	Result	Unit	Unit Reference Range	Test Name	Result	Unit	Unit Reference Range
WBC	5.95	10^3/µl	4.39 - 11.59	ALT	19	U/L	10 - 49
RBC	4.88	10^6/µl	4.14 - 5.37	AST	26	U/L	0 - 34
PLT	296	10^3/µL	152 - 383	Albumin	45	g/L	32 - 53
HGB	14.1	g/dL	12.2 - 15.9	ALP	279.8	U/L	46 - 116
HCT	40.3	%	36.4 - 47.2	T. BIL	2.05	mg/dL	0.3 - 1.2
MCV	82.6	fL	77.2 - 95.7	D. BIL	0.44	mg/dL	0 - 0.5
NE#	2.47	10^3/µL	2.04 - 7.54	I. BIL	1.61	mg/dL	0.3-1.0
LY#	2.95	10^3/µL	1.21 - 3.77	GGT	2	U/L	0 - 38
MO#	0.34	10^3/µL	0.26 - 0.94	HBs Ag	0.22	S/CO	EG<1.00-GRZ >=1.00
EO#	0.14	10^3/µL	0.02 - 0.5	Anti HCV	0.10	S/CO	<1
BA#	0.05	10^3/µL	0.01 - 0.07	Anti-HAV IgM	0.10	S/CO	>0.8
				Anti-HBc IgM	0.14		>0.8
				Anti-HBc IgG	0.08		0.79 - 1

WBC: White Blood Cell; NE: Neutrophil; LY: Lymphocyte; MO: Monocyte; EO: Eosinophil; BA: Basophil; RBC: Red Blood Cell; HGB. Hemoglobin; PLT: Platelet; HCT: Hematocrit; MCV: Mean corpuscular volume; MCHC:Mean cell heamoglobine concentration; ALP: alkaline phosphatase; ALT: alanine aminotransferase; AST:Aspartate aminotransferase; BUN; Blood urea nitrogen; GGT: Gamma-glutamyltransferase; TBIL: Total bilirubin; DBIL: Direct bilirubin; HBs Ag: Hepatitis B surface antigen; Anti HCV: Hepatitis C virus Antibodies; Anti-HAV IgM: Hepatitis A virüs immunoglobulin M Antibodies; Anti-HBc IgM: Immunoglobulin M antibody to hepatitis B core antigen; Anti-HBc IgG: Immunoglobulin G antibody to hepatitis B core antigen; I.BIL:Indirect Bilirubin)

Table 2. The results of the blood test analyses of Case 2.

Test Name	Result	Unit	Unit Reference Range	Test Name	Result	Unit	Unit Reference Range
WBC	7.49	10^3/µL	4.39 - 11.59	ALT	15	U/L	10 - 49
RBC	5.43	10^6/µl	4.14 - 5.37	AST	13	U/L	0 - 34
PLT	263	10^3/µL	152 - 383	GGT	7	U/L	0 - 38
NE#	3.32	10^3/µL	2.04 - 7.54	ALP	73.7	U/L	46 - 116
MO#	0.49	10^3/µL	0.26 - 0.94	T. BIL	1.47	mg/dL	0.3 - 1.2
MCV	89.0	fL	77.2 - 95.7	D. BIL	0.55	mg/dL	0 - 0.5
LY#	3.37	10^3/µL	1.21 - 3.77	I. BIL	0.92	mg/dL	0.3-1.0
HGB	16.5	g/dL	12.2 - 15.9	Albumin	45	g/L	32 - 53
HCT	48.3	%	36.4 - 47.2	HBs Ag	0.34	S/CO	EG<1.00-GRZ >=1.00
EO#	0.25	10^3/µL	0.02 - 0.5	Anti-HBc IgM	0.11		>0.8
BA#	0.06	10^3/µL	0.01 - 0.07	Anti-HBc IgG	0.06		0.79 - 1
				Anti HCV	0.06	S/CO	<1

WBC: White Blood Cell; NE: Neutrophil; LY: Lymphocyte; MO: Monocyte; EO: Eosinophil; BA: Basophil; RBC: Red Blood Cell; HGB. Hemoglobin; PLT: Platelet; HCT: Hematocrit; MCV: Mean corpuscular volume; MCHC:Mean cell heamoglobine concentration; ALP: alkaline phosphatase; ALT: alanine aminotransferase; AST:Aspartate aminotransferase; BUN; Blood urea nitrogen; GGT: Gamma-glutamyltransferase; TBIL: Total bilirubin; DBIL: Direct bilirubin; HBs Ag: Hepatitis B surface antigen; Anti HCV: Hepatitis C virus Antibodies; Anti-HAV IgM: Hepatitis A virüs immunoglobulin M Antibodies; Anti-HBc IgM: Immunoglobulin M antibody to hepatitis B core antigen; Anti-HBc IgG: Immunoglobulin G antibody to hepatitis B core antigen; I.BIL:Indirect Bilirubin)

3. Discussion

As the first point of contact for the patient, the family physician has to consider the profit and loss of the patient's health and the healthcare system. Therefore, the family physician is the first and most important point of contact both in the provision of health care and in the cost-effective management of healthcare services.

In line with the core competence of being community oriented, the family doctor adopts the basic principle of providing an effective and appropriate health service. The main aim is to prevent the conflict that can arise between the concerns, wishes and needs of the patient who comes to him and the needs and wishes of the whole health system and, therefore, of society. At this point, the family doctor can solve the problem as a first step with the confidence he/she has in the patient. In cases where it is necessary to turn to a secondary or tertiary health institution, he/she can provide a more effective health service by taking on the role of patient advocate. Therefore, family medicine plays a very important role in the referral to health centers after the first point of contact.

Failure to address the patient's concerns, failure to demonstrate correct management in the treatment and/or follow-up process of the disease may result in referral to higher level or outpatient application. This can lead to a cumulative burden on the healthcare system and a general impact on society in terms of healthcare utilization. Taking all this into account, the core competence of primary care management becomes very important. And to put this principle into practice, in addition to the clinical skills, knowledge and experience of the doctor, managerial skills in process management are also important.

The two cases mentioned in this article were from the same family, and the 18-year-old male patient was identified by history taking. History taking is of great importance in family medicine as in all other specialties. The history revealed that the jaundice occurred during periods of intense stress, insomnia and starvation, and then remitted. In accordance with the principle of primary care management of the discipline of family medicine, Gilbert's syndrome was diagnosed in patients with indirect hyperbilirubinemia without evidence of liver disease or hemolysis. Patients' concerns were allayed, they were told that they did not need to be referred to higher level hospitals, and they were given detailed information about the condition. In this way, the effective management of primary care and the effective use of its facilities solved a problem that is common in society but can cause anxiety for both patients and doctors.

4. Conclusion

In our country, the process of organizing primary health care is not yet complete. The main role in the completion of this process is played by family doctors. More effective use of primary health care can provide more qualified health care with a higher satisfaction rate in solving patients' health problems. In this way, a contribution can be made to the process of organizing primary health care services.

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Ethics

Informed Consent: The authors declared that informed consent form was signed by the patient.

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