İstanbul Kent Üniversitesi Sağlık Bilimleri Dergisi (IKUSBD)
Istanbul Kent University Journal of Health Science (IKUJHS)
Original Article/ Araştırma Makalesi e-ISSN: 2822-6615

DİŞ HEKİMLİĞİ ÖĞRENCİLERİNDE NADİR HASTALIK EĞİTİMİNDE BİLGİ-HAZIRLIK UYUMSUZLUĞU

A KNOWLEDGE–PREPAREDNESS GAP IN RARE-DISEASE EDUCATION AMONG DENTAL STUDENTS

Ezgi Gizem BERKAY[1*], Ata Berk ANGI[2]
[1]İstanbul Kent ,Üniversitesi, ezgi.berkay@kent.edu.tr, : 0000-0002-1967-705X [*]Sorumlu Yazar
[2] İstanbul Kent Üniversitesi, ataberkangi@gmail.com, 0009-0002-3297-1920

Geliş tarihi/Received : 16.06.2026 Kabul tarihi/Accepted: 22.08.2026 Yayın tarihi/Published: 24.09.2026

ÖZET

Giriş: Oral ve kraniyofasiyal bulgular nadir hastalıkların (NH) sıklıkla ilk ya da tek belirtisidir; buna karşın diş hekimliği müfredatlarında bu konuya nadiren yer verilmektedir.

Amaç: Bu çalışmada, diş hekimliği öğrencilerinin NH farkındalığı ve bir seçmeli dersin bu farkındalık üzerindeki etkisi değerlendirilmiştir.

Gereç ve Yöntem: Tek merkezli, tek kollu, ön test-son test tasarımına sahip bu çalışmada, 14 haftalık "Nadir Hastalıklar ve Diş Hekimliği" seçmeli dersine kayıtlı öğrenciler, Kühne ve ark.'nın anketinden uyarlanan anketi ders öncesinde ve sonrasında (Mart–Haziran 2023) yanıtlamıştır. Sonuç ölçütleri bileşik bilgi puanı, özbildirime dayalı hazır olma algısı, isimlendirilmiş NH'lerin tanınması ve NH veritabanlarına ilişkin farkındalıktır. Bilgi puanındaki değişim Wilcoxon işaretli sıra testi, madde düzeyindeki değişimler ise McNemar testi (Holm–Bonferroni düzeltmeli) ile değerlendirilmiştir.

Bulgular: Derse kayıtlı 119 öğrenciden 55'i her iki değerlendirmeyi de tamamlamıştır (%46,2; %63,6 kadın; yaş ortalaması 22,6). Bileşik bilgi puanı 6 üzerinden 1,11±1,24'ten 3,96±1,30'a yükselmiştir ($Z=-6{,}30$, $p<0{,}001$, $r=0{,}85$). Özbildirime dayalı hazır olma algısı sınırlı düzeyde iyileşmiş, sendromik veya genetik NH'lerin tanınma oranı ders sonrasında da düşük kalmıştır.

Tartışma: Bulgular, kısa süreli bir seçmeli dersin NH bilgisini belirgin biçimde artırdığını, ancak öğrencilerin klinik hazır olma algısını veya hastalık tanıma düzeyini iyileştirmede yetersiz kaldığını göstermektedir.

Sonuç: Kısa süreli bir seçmeli ders NH bilgisini artırmakla birlikte, algılanan klinik hazır olma durumunu artırmada tek başına yeterli değildir.

Öneriler: NH içeriğinin tek bir seçmeli derse sıkıştırılmak yerine klinik derslere boylamsal biçimde entegre edilmesi önerilmektedir.

Anahtar Kelimeler: *Nadir hastalıklar, diş hekimliği eğitimi, eğitim müdahalesi*

İstanbul Kent Üniversitesi Sağlık Bilimleri Dergisi (IKUSBD)
Istanbul Kent University Journal of Health Science (IKUJHS)
Original Article/ Araştırma Makalesi e-ISSN: 2822-6615

ABSTRACT

Introduction: Oral and craniofacial signs are frequently the presenting or sole manifestation of rare diseases (RD), yet dental curricula rarely address them.

Aim: This study assessed dental students' RD awareness and the impact of a dedicated elective course on that awareness.

Materials and Methods: In this single-centre, single-arm pre-post study, students enrolled in the 14-week elective "Rare Diseases and Dentistry" completed an identical questionnaire, adapted from Kühne et al., before and after the course (March-June 2023). Outcomes were a composite factual knowledge score, self-perceived preparedness, recognition of named RDs, and awareness of RD databases. The pre-post change in the knowledge score was tested using the Wilcoxon signed-rank test, and item-level changes using McNemar's test (Holm-Bonferroni-corrected).

Results: Of 119 enrolled students, 55 completed both assessments (46.2%; 63.6% female; mean age 22.6 years). The composite knowledge score rose from 1.11±1.24 to 3.96±1.30 of 6 ($Z=-6.30$, $p<0.001$, $r=0.85$). Self-rated preparedness improved only modestly, and recognition of most syndromic or genetic RDs remained low after the course.

Discussion: The findings indicate that a short elective course markedly increases factual RD knowledge but is insufficient to improve students' perceived clinical preparedness or disease-recognition ability.

Conclusion: A short elective course increases RD knowledge but is not, by itself, sufficient to improve perceived clinical preparedness.

Recommendations: RD content should be embedded longitudinally across clinical courses rather than confined to a single elective.

Keywords: *Rare diseases; dental education; educational intervention*

INTRODUCTION

Rare diseases (RD), which are increasingly important not only in medical genetics but also across nearly all medical fields, are defined as diseases with a prevalence of less than 1 in 2,000 and are recognised within the European Union and Türkiye (Moliner and Waligora, 2017). This definition applies to diseases affecting fewer than 1 in 200,000 individuals in the United States and fewer than 1 in 2,500 in Japan (Mizoguchi et al., 2016; Marwaha et al., 2022). Up to 80% of rare diseases are genetic in origin, and between 6,000 and 8,000 have been identified worldwide (EURORDIS, 2023).

Oral and dental findings can indicate many rare diseases and may be the only signs in mild cases, so it is important for dentists to be aware of RD and able to recognize it. These patients undergo a diagnostic process that often involves extensive, costly tests across multiple institutions, and they are frequently underdiagnosed or misdiagnosed. The average time to receive an accurate diagnosis for a rare disease is about 4-5 years, and in some cases, it can take longer than 10 years. A proper diagnosis is essential because it guides the physician, patient, and family by ensuring appropriate treatment, avoiding ineffective therapies, predicting prognosis and recurrence risk, and helping prevent potential complications.

In this context, it is crucial for all healthcare professionals to be aware of RD and to understand the steps required to recognize it, refer patients to qualified institutions, and diagnose the most common genetic disorders. The Ministry of Health of Türkiye prepared “The Rare Diseases Health Strategy Document and Action Plan,” which prioritizes healthcare personnel and students for future directions (T.C. Sağlık Bakanlığı, 2023), yet dental curricula rarely address RD in detail.

In this study, the questionnaire method was used to assess dentistry students’ (n=55) awareness of rare diseases and the impact of an elective course, “The Rare Diseases and Dentistry,” on this awareness. Using a single-arm pre-post (quasi-experimental, uncontrolled) design, the same cohort was assessed before and after the 14-week course. We hypothesised that the course would increase factual knowledge of RD; as secondary aims, we examined whether it also changed self-perceived clinical preparedness, recognition of named RDs, and awareness of RD information resources.

METHOD

This single-arm pre-post study evaluated dental students’ awareness of RD and examined the influence of education on this topic. To achieve this, the questionnaire originally developed by Kühne et al. for dentists in Germany was adapted for dental students in Türkiye (Kühne et al., 2020). Students from the [institution masked for peer review] Faculty of Dentistry who had started their clinical training, enrolled in the “Rare Diseases and Dentistry” course, and agreed to participate completed the questionnaire at the beginning and end of the semester via a Google Forms link. Pre- and post-course

responses were linked at the individual level using a self-generated anonymous code, enabling paired analysis. This method allowed for an assessment of educational effects. The same questions appeared in both the initial and follow-up surveys. The first page of the survey provided information about the study and a consent form. The first survey was conducted from March 1 to March 15, 2023. The second survey was available from June 1 to June 8, 2023. Participants were excluded if they submitted incomplete questionnaires.

The questionnaire included 22 questions divided into five sections. Five questions collected general demographic information; seven assessed knowledge of rare diseases; one evaluated these diseases, including 17 disease names; three covered educational status; and six explored information needs (Supplementary 1). The study is reported in accordance with the STROBE statement for observational studies (Supplementary 4). The German questionnaire of Kühne et al. was translated into Turkish and independently back-translated by two bilingual researchers, and its content was reviewed by specialists in medical genetics and dental education before administration; the internal consistency of the knowledge score was modest at baseline (Cronbach's α/KR-20 = 0.61). The licensing status of this instrument is confirmed in Appendix 1. Data were analysed with IBM SPSS Statistics v25. Categorical variables are summarised as frequencies and percentages, and the knowledge score as mean ± standard deviation. A composite knowledge score was computed for each participant by summing correct responses to the factual knowledge items. The primary score was based on the six factual items; the item asking whether RDs constitute a public health problem was excluded because it assesses opinion rather than knowledge, and the originally computed seven-item score is reported in a sensitivity analysis (Supplementary 2). Recognition of named RDs (0-17) and awareness of RD databases were each summed into a score and compared pre-post with the Wilcoxon signed-rank test. Because the score is a bounded count and its paired difference deviated from normality (Lilliefors $p<0.05$), the Wilcoxon signed-rank test served as the primary test of the pre–post change, with the paired-samples t-test reported alongside for comparison; effect sizes are reported as $r=Z/\sqrt{N}$ and Cohen's d, respectively. Changes in the proportion of correct responses to each individual knowledge item were tested with McNemar's test, with Holm-Bonferroni correction for multiple comparisons. Two-sided $p<0.05$ was considered significant.

This study was approved by the [institution masked for peer review] Ethics Committee (approval no. 2023-04) and was conducted in accordance with the Declaration of Helsinki (as revised in 2013). All participants provided informed consent prior to completing the questionnaire.

This study has some limitations that should be considered when interpreting its results. The single-arm, uncontrolled pre-post design cannot separate the contribution of the elective from maturation, repeated testing, or secular trends. The study was conducted at a single centre with a modest,

self-selected sample (55 of 119 enrolled students completed both assessments), which may limit generalisability. The questionnaire's internal consistency was only moderate at baseline, and preparedness was self-reported rather than objectively measured. Only the immediate post-course response was assessed, so knowledge retention remains unknown. The study also did not incorporate institutional examination or assessment data for the elective course, which would have allowed a more objective evaluation of knowledge acquisition than the self-administered survey used here. Future evaluations of similar interventions should include validated institutional assessment data alongside self-report measures. Baseline characteristics of students who did not complete both assessments were not compared with those of completers, so the extent of attrition-related bias cannot be fully excluded. The list of named RDs used to assess recognition included conditions of varying rarity and clinical presentation, and their selection was not based on a formal, prespecified case-mix or severity classification; this may limit the construct validity of the recognition score and should be addressed with a more systematically developed instrument in future work.

FINDINGS and DISCUSSION

Of the 119 students enrolled in the course, 75 completed the first questionnaire, and 60 completed the second. Of these, 55 students completed both surveys and could be matched at the individual level; these paired records formed the analysis sample (46.2% (n=55) of those enrolled). Among these 55 participants, 63.6% (n=35) were female, and 36.4% (n=20) were male, with a mean age of 22.6 years. While 20 students beginning clinical semester courses were in their third year of university, 35 were in their fourth year. Only 16 respondents (29.1%) reported having met an individual with an RD, and 8 (14.5%) had a family member or close contact diagnosed with a rare disease. Additionally, 87.3% (n=48) reported having heard of the concept of RD before the course.

At the end of the education period, questions regarding the lessons' contribution to awareness were evaluated, and the response rates are shown in the table below (Table 1). The composite knowledge score increased from 1.11±1.24 before the course to 3.96±1.30 afterwards (maximum 6). The Wilcoxon signed-rank test confirmed a significant improvement ($Z=-6.30$, $p<0.001$, $r=0.85$): scores were higher after the course in 52 of 55 participants, unchanged in 3, and lower in none; the paired-samples t-test was concordant ($t(54)=13.11$, $p<0.001$, $d=1.77$) (Supplementary 2). The result was materially unchanged in a sensitivity analysis using the original seven-item score ($Z=-6.30$, $p<0.001$). At the item level, the largest gains in correct responses were for the incidence of RD (10.9% (n=6) to 76.4% (n=42)), the most affected age group (25.4% (n=14) to 94.5% (n=52)), and the proportion of RD that are genetic (10.9% (n=6) to 70.9% (n=39)); the most common cause also rose (50.9% (n=28) to 85.5% (n=47)), whereas the estimated number of identified RD (10.9% (n=6) to 43.7% (n=24)) and the number of patients in Türkiye (3.6% (n=2) to 29.1% (n=16)) remained below 50% after the course. After Holm-

Bonferroni correction, all six factual items showed a significant pre–post increase (all adjusted p<0.05; Table 1).

Table 1. *The proportions of the answers given by the participants to the questions before and after the lessons*

Question / Response option	1st response, % (n)	2nd response, % (n)
What is the incidence of rare diseases?		
1/1000	3.6% (n=2)	0% (n=0)
1/2000	10.9% (n=6)	76.4% (n=42)
1/3000	0% (n=0)	3.6% (n=2)
1/4000	1.8% (n=1)	0% (n=0)
1/5000	7.3% (n=4)	3.6% (n=2)
1/10,000	1.8% (n=1)	1.8% (n=1)
1/100,000	18.2% (n=10)	7.3% (n=4)
I don't know	56.4% (n=31)	7.3% (n=4)
How many rare diseases have been identified?		
100-500	3.6% (n=2)	1.8% (n=1)
1000-2000	5.5% (n=3)	14.6% (n=8)
3000-5000	1.8% (n=1)	9% (n=5)
6000-8000	10.9% (n=6)	43.7% (n=24)
9000-10,000	1.8% (n=1)	3.6% (n=2)
More than 10,000	1.8% (n=1)	14.6% (n=8)
I don't know	74.6% (n=41)	12.7% (n=7)
Rare diseases are most common in which age group?		
Newborn and childhood	25.4% (n=14)	94.5% (n=52)
Puberty	5.5% (n=3)	0% (n=0)
Adulthood	5.5% (n=3)	0% (n=0)
Common in all ages	12.7% (n=7)	0% (n=0)
I don't know	50.9% (n=28)	5.5% (n=3)
How many individuals are diagnosed with rare diseases in Türkiye?		
1000-5000	3.6% (n=2)	12.7% (n=7)
5000-10,000	3.6% (n=2)	14.6% (n=8)
10,000-15,000	3.6% (n=2)	7.3% (n=4)
15,000-20,000	3.6% (n=2)	3.6% (n=2)

25,000-35,000	1.8% (n=1)	0% (n=0)
50,000-100,000	5.5% (n=3)	3.6% (n=2)
More than 100,000	3.6% (n=2)	29.1% (n=16)
I don't know	74.6% (n=41)	29.1% (n=16)
What is the most common cause of rare diseases?		
Autoimmunity	12.7% (n=7)	12.7% (n=7)
Mitochondrial disorders	1.8% (n=1)	0% (n=0)
Genetics	50.9% (n=28)	85.5% (n=47)
Infections	1.8% (n=1)	0% (n=0)
Environmental factors	1.8% (n=1)	0% (n=0)
I don't know	30.9% (n=17)	1.8% (n=1)
Rare genetic diseases account for what percentage of all rare diseases?		
10%	1.8% (n=1)	3.6% (n=2)
20%	7.3% (n=4)	12.7% (n=7)
50%	14.6% (n=8)	1.8% (n=1)
80%	10.9% (n=6)	70.9% (n=39)
100%	0% (n=0)	0% (n=0)
I don't know	65.4% (n=36)	10.9% (n=6)
Do rare diseases constitute a significant public health problem?		
Definitely yes	36.3% (n=20)	40% (n=22)
Yes	40% (n=22)	43.6% (n=24)
No	5.5% (n=3)	7.3% (n=4)
Definitely no	0% (n=0)	1.8% (n=1)
I don't know	18.2% (n=10)	7.3% (n=4)

McNemar test with Holm-Bonferroni correction (six factual items; the public-health item was excluded as an opinion item). All items improved significantly: incidence, adjusted $p = 1.4\times10^{-9}$; number identified, $p = 4.6\times10^{-5}$; age group, $p = 4.4\times10^{-11}$; number in Türkiye, $p = 5.5\times10^{-4}$; most common cause, $p = 5.5\times10^{-4}$; genetic proportion, $p = 4.3\times10^{-9}$.

Dentistry students' knowledge and perspectives on the professional examination and treatment of individuals with RD are presented in Table 2. Responses to the question "Which of the rare diseases seen in our country have you heard of?" evaluate awareness of RD and related databases and education needs (Tables 3 and 4). The recognition score increased modestly, from 7.3±3.7 to 8.9±4.3 of 17 ($Z=-2.10$, $p=0.036$, $r=0.28$), with wide individual variation (29 students recognised more diseases, 19 fewer, and 7 unchanged). In contrast, awareness of RD databases rose more substantially, from 0.6±0.8 to 2.0±1.6 ($Z=-4.97$, $p<0.001$, $r=0.67$).

Table 2. *Dentistry Students' knowledge needs and their thoughts on the examination and treatment of individuals with rare diseases in their professional lives*

Question / Response option	1st response, % (n)	2nd response, % (n)
Are you ready to treat an individual diagnosed with a rare disease?		
Definitely	3.6% (n=2)	9.1% (n=5)
I'm a little ready	14.6% (n=8)	32.7% (n=18)
I'm not quite ready	29.1% (n=16)	38.2% (n=21)
I'm definitely not ready	41.8% (n=23)	16.4% (n=9)
I'm undecided	10.9% (n=6)	3.6% (n=2)
Do you think that rare diseases have a place and importance in dentistry?		
Yes	81.8% (n=45)	81.8% (n=45)
Partially	9.1% (n=5)	14.6% (n=8)
No	0% (n=0)	1.8% (n=1)
No idea	9.1% (n=5)	1.8% (n=1)
Do you think your knowledge about rare diseases is sufficient?		
Yes	0% (n=0)	12.7% (n=7)
Partially	18.2% (n=10)	69.1% (n=38)
No	81.8% (n=45)	18.2% (n=10)

It is crucial for dentists to be aware of RD and to recognize it, as oral and dental findings can indicate many diseases and may be the only signs in mild cases. Studies assessing awareness of this topic have been conducted with dentists and students from various health fields. The common conclusion from numerous studies is that all health professionals need more education and collaboration regarding RD (Kühne et al., 2020; Domaradzki and Walkowiak, 2019; Vandeborne et al., 2019; Walkowiak and Domaradzki, 2020; Li et al., 2021; Mijiritsky et al., 2021; Walkowiak and Domaradzki, 2021; Benz et al., 2022). Similar needs have been reported among physicians in Spain (Ramalle-Gómara et al., 2020). Our current knowledge indicates that no survey has been conducted or published in Türkiye on this subject. To our knowledge, few studies have used a pre-post design to evaluate the effects of adding an elective rare-disease course to the undergraduate dental curriculum on student awareness.

Table 3. *Responds: "Which of the rare diseases seen in our country have you heard of?"*

Disease	1st response (n)	2nd response (n)
Sickle cell anaemia	87.3% (n=48)	89% (n=49)
Cystic Fibrosis	69% (n=38)	74.5% (n=41)
Acromegalia	69% (n=38)	80% (n=44)
Haemophilia	87.3% (n=48)	92.7% (n=51)
Down Syndrome	90.9% (n=50)	90.9% (n=50)
Niemann-Pick Disease	18.2% (n=10)	29% (n=16)
Progeria	7.3% (n=4)	25.4% (n=14)
Neurofibromatosis	23.6% (n=13)	34.5% (n=19)
Cerebral Palsy	43.6% (n=24)	54.5% (n=30)
Huntington Disease	52.7% (n=29)	60% (n=33)
Mucopolysaccharidosis	10.9% (n=6)	27.2% (n=15)
Crohn Disease	36.3% (n=20)	60% (n=33)
Pompe Disease	1.8% (n=1)	20% (n=11)
Fragile X syndrome	45.4% (n=25)	50.9% (n=28)
Marfan Syndrome	9.1% (n=5)	29.1% (n=16)
Osteogenesis imperfecta	47.3% (n=26)	58.2% (n=32)
Phenilketonuria	30.9% (n=17)	43.6% (n=24)

Table 4. *Awareness of rare-disease-related databases before and after the lessons*

Database	1st Respond (n)	2nd Respond (n)
NORD	7.3% (n=4)	25.4% (n=14)
EURORDIS	0% (n=0)	20% (n=11)
Orphanet	9.1% (n=5)	40% (n=22)
OMIM	3.6% (n=2)	34.5% (n=19)
PubMed	41.8% (n=23)	76.4% (n=42)
I don't know	54.5% (n=30)	18.2% (n=10)

This intervention can also be considered within the framework of competency-based dental education, in which an elective course is expected to align with broader program competencies and graduate outcome frameworks, such as national core curricula or the profile and competences defined by the Association for Dental Education in Europe (ADEE) (Cowpe et al., 2010). Framed this way, the gap between factual knowledge and perceived preparedness observed in this study suggests that the elective addressed cognitive learning objectives more effectively than the clinical, diagnostic, and referral competencies expected of a dental graduate.

The Rare Diseases Report, published in 2019 by the Turkish Institute of Public Health and Chronic Diseases, an institute affiliated with the Health Institutes of Türkiye (TÜSEB), reports an average prevalence of RD in Türkiye of 38 per 100,000 people. It estimates that around 5 million individuals in the country have RD, including approximately 30,300 with ultra-rare disorders (Satman

et al., 2019). According to 2023 data from the Turkish Statistical Institute (TÜİK), Türkiye's population is 85,279,553, accounting for 1.1% of the global population (TÜİK, 2023). Given that about 350 million people worldwide have rare diseases, it is estimated that roughly 3,850,000 individuals in Türkiye also have RD. However, this number is below 5 million and is believed to be associated with highly prevalent consanguineous marriages. Data from 2018 shows that 24.1% of marriages were consanguineous, but this rate dropped to 8.3% for new marriages in 2022 (Hacettepe Üniversitesi, 2019; TÜİK, 2023). Given that approximately 15% of RDs present with orofacial manifestations, it can be inferred that approximately 52,500,000 patients worldwide and 750,000 in Türkiye may also require dental care.

Despite these high rates, specialized physicians, nurses, and dentists state that their knowledge in this area is insufficient. Recognizing that even medical faculties have deficiencies in RD education, it is not surprising that dentistry falls short in this regard (Vandeborne et al., 2019; Li et al., 2021). In this context, in addition to encountering individuals with RD at any stage of their professional life, it is almost inevitable that they take part in diagnosing and treating individuals with RD and orofacial involvement. The diagnosis and treatment of these individuals will surely be delayed due to insufficient knowledge and awareness today. Considering that some RDs' mild forms may occur only with oral and/or dental involvement (ex., Cleidocranial dysplasia), it is possible that they cannot be diagnosed at all. For this reason, it is necessary to develop a roadmap that ensures professionals trained across all health fields are comprehensively informed about common conditions and RD, as well as the workflow they should follow when examining a rare disease.

Although the European Union Council stated in 2009 that all healthcare professionals should actively participate in early diagnosis and treatment (Moliner and Waligora, 2017), survey studies have shown that significant progress has not been achieved. In a study involving 5 RD specialists and 295 physicians in Belgium, physicians' basic knowledge of RD was poor: 22% of general practitioners and 15% of paediatricians were unaware of the sources they should consult for RD information. It was reported that 72% of paediatricians, who encounter it most often, have an average or below-average understanding of the subject, indicating a need for improved academic training (Vandeborne et al., 2019). Similarly, another study of 165 medical doctors in Poland found that knowledge of the aetiology, epidemiology, and frequency of RD, as well as differences between common and rare diseases and how to differentiate them, was lacking; 94.6% rated their knowledge as inadequate (Walkowiak and Domaradzki, 2021). In a subsequent study involving 9 RD specialists and 224 physicians in China, only 5.3% had an average or good understanding of RD (Li et al., 2021). Additionally, a survey of 346 medical students in Poland found that students reported insufficient understanding of rare diseases; they felt unprepared to manage such patients in their professional practice and expressed a need for more comprehensive training in this area (Domaradzki and Walkowiak, 2019).

To the best of our knowledge, three studies have been conducted with dentists: two in Germany and one in Israel. A 2021 study assessing the knowledge of dentists, orthodontists, periodontists, and oral and craniomaxillofacial surgeons regarding RD in Westphalia-Lippe, Germany, reported that specialist dentists working at universities had higher levels of knowledge than general dentists. Despite differences between the groups, knowledge levels were found to be lacking across all groups (Kühne et al., 2020). Additionally, a study involving 309 dentists in Germany concluded that expertise levels need improvement and that information about RD is essential to their daily routines (Benz et al., 2022). Another survey conducted in Israel (n=309) revealed that most information about RD is obtained from a few-day workshops, conferences, or lectures, and that dentists require more information (Mijiritsky et al., 2021).

The "2023-2027 Rare Diseases Health Strategy Document and Action Plan" has been published by the Ministry of Health in our country. Within this framework, it is planned to increase awareness among all healthcare professionals about RD and to promote active involvement in early diagnosis and treatment. Given the high prevalence of RD and the widespread practice of consanguineous marriages in Türkiye, this study aims to identify gaps in this area.

The content of the 14-week theoretical lecture is presented in the table below (Table 5). In addition to providing general information and raising awareness of RD, it aims to help students recognize anomalies and dysmorphic features in the orofacial region and consider the possibility of rare genetic syndromes or single-gene disorders. Based on these findings, another study focused on creating a database for RD, reporting that 15% of RD cases involved orofacial features, and more than 900 genetic syndromes included dental, oral, or maxillofacial manifestations (Hanisch et al., 2019). Although the level of awareness and knowledge about RD improved significantly following the elective, responses indicated that if they were preparing to diagnose and treat individuals with RD in their professional lives, their readiness changed from "I'm definitely not ready" (41.8% (n=23)) to "I'm not quite ready" (38.2% (n=21)). The knowledge they gained on this subject remained partial (69.1% (n=38)). This study highlights the need to include these topics in related courses (e.g., pedodontics, endodontics) and to provide more detailed information on genetic diseases to prepare students better to manage RD in their dental careers.

Table 5. *Course content according to the weekly programs*

Week	Topics Covered
1	Introduction to rare diseases
2	Diagnostic approaches in rare diseases
3	Therapy options for rare diseases
4	Patient organizations for rare diseases
5	Newborn screening, metabolic disorders, inborn errors in metabolism
6	Rare genetic syndromes
7	Rare immunodeficiencies
8	Rare malignancies
9	Rare neurological diseases
10	Rare diseases with oral component I
11	Rare diseases with oral component II
12	Rare diseases with craniofacial manifestations I
13	Rare diseases with craniofacial manifestations II
14	Case discussion

Another important finding concerns the recognition of individual RDs by name. Recognition increased for nearly all of the listed conditions (the overall recognition score rose from 7.3 to 8.9 of 17; $Z=-2.10$, $p=0.036$, $r=0.28$), with the largest gains for less familiar syndromic or genetic disorders (e.g., Pompe disease: 1.8% (n=1) to 20.0% (n=11); Marfan syndrome: 9.1% (n=5) to 29.1% (n=16); progeria: 7.3% (n=4) to 25.4% (n=14)). However, recognition of most of these conditions remained below one-third after the course, while already-familiar disorders such as Down syndrome and sickle cell anaemia were near the ceiling at both time points (Table 3). The course, therefore, broadened, but did not consolidate, recognition of genetic RDs. This result suggests that these lessons should be studied not only in “rare diseases” but also in related courses, and that the level of knowledge needs to be increased. In this context, one option is to have field experts teach information on genetic diseases by incorporating it into clinical courses through committee lectures, rather than in short-term classes (Supplementary 3). Although awareness of databases related to RD varies, this study did not assess whether participants use these tools or the extent of their knowledge of their contents. Therefore, the use of databases can be demonstrated in practice, and habits can be improved. A paired analysis of the ordinal preparedness responses in Table 2 (excluding undecided answers, N=48) confirmed a significant improvement (Wilcoxon $Z=-3.43$, $p<0.001$, $r=0.50$; improved in 27 of 48). Taken together, the effect size was largest for factual knowledge ($r=0.85$), intermediate for perceived preparedness ($r=0.50$), and smallest for

disease recognition (r=0.28), indicating that the course strengthened declarative knowledge more than the applied readiness and recognition skills expected of a dental graduate.

CONCLUSION AND RECOMMENDATIONS

To our knowledge, few studies have measured dentistry students' awareness of RD or evaluated the impact of a dedicated RD course; this study provides empirical evidence in this area. The data suggest that integrating rare and genetic disease content into healthcare education, alongside case-based discussions, may help improve future practitioners' ability to diagnose and manage individuals with RD during their professional practice.

Appendices

Appendix 1. Licensing status of the measurement instrument

The questionnaire used in this study was adapted from the instrument originally developed by Kühne et al. (2020) and published in the International Journal of Environmental Research and Public Health (MDPI). This article is licensed under the Creative Commons Attribution 4.0 International (CC BY 4.0) license, as stated on the publisher's article page; under this license, the article and its content may be freely reused, adapted, and redistributed for any purpose, including research and educational use, provided that the original work is properly cited, with no separate permission or license fee required. Critically, the questionnaire itself was published by the original authors as open-access Supplementary Material to that article ("Score S1: Questionnaire and Categories", freely downloadable from the publisher's website), under the same CC BY 4.0 license as the article; it is therefore a specifically confirmed, openly licensed research instrument, not merely inferred from the journal's general policy. This study cites the original instrument (Kühne et al., 2020) accordingly. The instrument was not reproduced verbatim: it was translated into Turkish, independently back-translated by two bilingual researchers, and adapted for a dental-student population, with its content reviewed by specialists in medical genetics and dental education before administration. The full set of adapted items is provided in Supplementary 1 for transparency.

REFERENCES

Benz, K., Trapp, R., Voss, M., Hanisch, M., Geisthoff, U., & Jackowski, J. (2022). 'Awareness and knowledge of rare diseases in German dentists, dental specialists and oral and maxillofacial surgeons: A country-wide survey', *Medicina*, *58*(8), 1114. https://doi.org/10.3390/medicina58081114

Cowpe, J., Plasschaert, A., Harzer, W., Vinkka-Puhakka, H., & Walmsley, A. D. (2010). 'Profile and competences for the graduating European dentist – update 2009', *European Journal of Dental Education*, *14*(4), pp. 193–202. https://doi.org/10.1111/j.1600-0579.2009.00609.x

Domaradzki, J., & Walkowiak, D. (2019). 'Medical students' knowledge and opinions about rare diseases: A case study from Poland', *Intractable & Rare Diseases Research*, *8*(4), pp. 252–259. https://doi.org/10.5582/irdr.2019.01099

EURORDIS. (n.d.). *What is a rare disease?* Retrieved August 8, 2023, from https://www.eurordis.org/information-support/what-is-a-rare-disease/

Hacettepe Üniversitesi Nüfus Etütleri Enstitüsü. (2019). *2018 Türkiye nüfus ve sağlık araştırması.* Hacettepe Üniversitesi. http://hdl.handle.net/11655/23356

Hanisch, M., Hoffmann, T., Bohner, L., Hanisch, L., Benz, K., Kleinheinz, J., & Jackowski, J. (2019). 'Rare diseases with periodontal manifestations', *International Journal of Environmental Research and Public Health*, *16*(5), 867. https://doi.org/10.3390/ijerph16050867

Kühne, A., Kleinheinz, J., Jackowski, J., Köppe, J., & Hanisch, M. (2020). 'Study to investigate the knowledge of rare diseases among dentists, orthodontists, periodontists, oral surgeons and craniomaxillofacial surgeons', *International Journal of Environmental Research and Public Health*, *18*(1), 139. https://doi.org/10.3390/ijerph18010139

Li, X., Zhang, X., Zhang, S., Lu, Z., Zhang, J., Zhou, J., Li, B., & Ou, L. (2021). 'Rare disease awareness and perspectives of physicians in China: A questionnaire-based study', *Orphanet Journal of Rare Diseases*, *16*(1), 171. https://doi.org/10.1186/s13023-021-01788-3

Marwaha, S., Knowles, J. W., & Ashley, E. A. (2022). 'A guide for the diagnosis of rare and undiagnosed disease: Beyond the exome', *Genome Medicine*, *14*(1), 23. https://doi.org/10.1186/s13073-022-01026-w

Mijiritsky, E., Dekel-Steinkeller, M., Peleg, O., Kleinman, S., Ianculovici, C., Shuster, A., Arbel, S., Ben-Ezra, M., & Shacham, M. (2021). ' Knowledge and associated factors about rare diseases among dentists in Israel: A cross sectional survey', *International Journal of Environmental Research and Public Health*, *18*(13), 6830. https://doi.org/10.3390/ijerph18136830

Mizoguchi, H., Yamanaka, T., & Kano, S. (2016). 'Research and drug development activities in rare diseases: Differences between Japan and Europe regarding influence of prevalence', *Drug Discovery Today*, *21*(10), pp. 1681–1689. https://doi.org/10.1016/j.drudis.2016.06.014

Moliner, A. M., & Waligora, J. (2017). The European Union policy in the field of rare diseases. In M. Posada De La Paz, D. Taruscio, & S. C. Groft (Eds.), *Rare diseases epidemiology: Update and overview* (Vol. 1031, pp. 561–587). Springer International Publishing. https://doi.org/10.1007/978-3-319-67144-4_30

Ramalle-Gómara, E., Domínguez-Garrido, E., Gómez-Eguílaz, M., Marzo-Sola, M. E., Ramón-Trapero, J. L., & Gil-de-Gómez, J. (2020). 'Education and information needs for physicians about rare diseases in Spain', *Orphanet Journal of Rare Diseases*, *15*(1), 18. https://doi.org/10.1186/s13023-019-1285-0

Satman, İ., Güdük, Ö., Yemenici, M., & Ertürk, N. (2019). *Nadir hastalıklar raporu.* Türkiye Halk Sağlığı ve Kronik Hastalıklar Enstitüsü (TÜHKE), TÜSEB. https://www.tuseb.gov.tr/tuhke/uploads/genel/files/haberler/nadir_hastaliklar_raporu.pd

T.C. Sağlık Bakanlığı. (2023). *2023–2027 nadir hastalıklar sağlık strateji belgesi ve eylem planı.* https://shgm.saglik.gov.tr/TR-101118/2023---2027-nadir-hastaliklar-saglik-strateji-belgesi-ve-eylem-plani.html

TÜİK. (2023). *Dünya nüfus günü, 2023.* Retrieved August 8, 2023, from https://data.tuik.gov.tr/Bulten/Index?p=Dunya-Nufus-Gunu-2023-49688

Vandeborne, L., Van Overbeeke, E., Dooms, M., De Beleyr, B., & Huys, I. (2019). 'Information needs of physicians regarding the diagnosis of rare diseases: A questionnaire-based study in Belgium', *Orphanet Journal of Rare Diseases*, *14*(1), 99. https://doi.org/10.1186/s13023-019-1075-8

Walkowiak, D., & Domaradzki, J. (2020). 'Needs assessment study of rare diseases education for nurses and nursing students in Poland', *Orphanet Journal of Rare Diseases*, *15*(1), 167. https://doi.org/10.1186/s13023-020-01432-6

Walkowiak, D., & Domaradzki, J. (2021). 'Are rare diseases overlooked by medical education? Awareness of rare diseases among physicians in Poland: An explanatory study', *Orphanet Journal of Rare Diseases*, *16*(1), 400. https://doi.org/10.1186/s13023-021-02023-9