

Sveikatos priežiūros specialistų profesinės kvalifikacijos tobulinimo kursas
„Daugiadisciplinė sergančiųjų retomis ligomis sveikatos priežiūra“

Klausytojai: visų profesinių kvalifikacijų gydytojai.

Trukmė: viso 40 akad. val.

Kursų turinys:

- 1) Kursų pristatymas. Bazinių žinių anketinė apklausa. Rezultatų aptarimas (1 akad. val.);
- 2) Paskaitų - 9 (18 akad. val.);
- 3) Seminarų – 17 (18 akad. val.);
- 4) Žinių patikrinimas. Rezultatų įvertinimas ir aptarimas. Kursų apibendrinimas (3 akad. val.).

Kursas bus užskaitytas ir pažymėjimas išduotas kursų dalyviams, kurie išklausė visą kursą ir aktyviai dalyvavo baigiamojoje diskusijoje (uždavė bent 1 klausimą) dėstytojams.

Parengė: doc. Birutė Tumienė, prof. Algirdas Utkus.

Kurso programos turinys

Temos pavadinimas	Užsiėmimo tipas	Trukmė (akad. val.)
Kursų pristatymas	Diskusija, apklausa	1
Retų ir genetinių ligų epidemiologija ir klasifikacija	Paskaita	2
Retų ligų srities iššūkiai ir jų sprendimo būdai	Paskaita	2
Retų ligų diagnostika	Paskaita	2
Specializuotas retų ligų gydymas	Paskaita	2
Sergančiųjų retomis ligomis integruotos priežiūros principai	Paskaita	2
Genetinės ir retos ligos slaugytojo praktikoje	Paskaita	2
Genetiko vaidmuo daugiadisciplinėje retų ligų priežiūroje	Paskaita	2
Retų ligų prevencija	Paskaita	2
Moksliniai retų ligų tyrimai	Paskaita	2
Paciente ir šeimos stiprinimas	Seminaras	2
Bioetiniai ir teisiniai klausimai	Seminaras	2
Daugiadisciplinė sergančiųjų paveldimomis medžiagų apykaitos ligomis sveikatos priežiūra	Seminaras	1
Daugiadisciplinė sergančiųjų retomis neurologinėmis ligomis sveikatos priežiūra	Seminaras	1
Daugiadisciplinė sergančiųjų retomis nervų ir raumenų ligomis sveikatos priežiūra	Seminaras	1
Daugiadisciplinė sergančiųjų retomis epilepsijomis sveikatos priežiūra	Seminaras	1
Daugiadisciplinė pacientų su retais klausos sutrikimais sveikatos priežiūra	Seminaras	1
Daugiadisciplinė sergančiųjų retomis akių ligomis sveikatos priežiūra	Seminaras	1
Daugiadisciplinė pacientų su įgimtomis kraujagyslių anomalijomis sveikatos priežiūra	Seminaras	1
Daugiadisciplinė sergančiųjų retomis endokrininės sistemos ligomis sveikatos priežiūra	Seminaras	1
Daugiadisciplinė pacientų su raidos anomalijomis ir intelektine negalia sveikatos priežiūra	Seminaras	1

Daugiadisciplinė sergančiųjų retomis širdies ligomis sveikatos priežiūra	Seminaras	1
Daugiadisciplinė sergančiųjų retomis inkstų ligomis sveikatos priežiūra	Seminaras	1
Daugiadisciplinė sergančiųjų retomis plaučių ligomis sveikatos priežiūra	Seminaras	1
Daugiadisciplinė sergančiųjų retomis odos ligomis sveikatos priežiūra	Seminaras	1
Daugiadisciplinė sergančiųjų retomis skeleto ir jungiamojo audinio ligomis sveikatos priežiūra	Seminaras	1
Žinių patikrinimas. Rezultatų įvertinimas. Kursų apibendrinimas.	Diskusija, apklausa	3
	Viso:	40

Retų ir genetinių ligų epidemiologija ir klasifikacija

Paskaita. 2 val. Retų ligų apibréžimas. Retų ligų kodavimas. Retų ir genetinių ligų klasifikacijos ir ontologija. Retų ligų epidemiologinių tyrimų svarba. Retų ir genetinių ligų dažnis bendroje populiacijoje ir įvairiose pacientų populiacijose pagal amžiaus grupes, sveikatos priežiūros paslaugas, medicinos sritis. Sergamumas ir mirtingumas. Retų ir genetinių ligų ekonominis poveikis. Medicininių intervencijų ekonominis poveikis.

Rekomenduojama literatūra.

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Retų ligų srities iššūkiai ir jų sprendimo būdai

Paskaita. 2 val. Neatliepti sergančiųjų retomis ligomis ir jų artimųjų poreikiai ir poveikis gyvenimo kokybei bei psichologinei sveikatai. Retų ir genetinių ligų diagnostikos iššūkiai, nediagnozuotos ligos. Retų ir genetinių ligų gydymo iššūkiai, retieji vaistai. Retų ligų ilgalaikės priežiūros iššūkiai: paslaugų koordinavimas ir atvejo vadyba, visapusiška, integruota sveikatos priežiūra, daugiadisciplinė priežiūra, pacientų keliai. Paciento ir sveikatos priežiūros specialisto komunikavimo problemas. Informacijos ir žinių stoka retų ligų srityje. Geografiniai netolygumai retų ligų srityje. Priemonės retų ligų srities iššūkiams spręsti.

Rekomenduojama literatūra.

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Retų ligų diagnostika

Paskaita. 2 val. Etiologinės retos ligos diagnozės nustatymo svarba pacientui, šeimai, visuomenei. Pirminės retos ligos diagnostika: svarbiausi simptomai, požymiai ir anamnezės duomenys įvairiose ligų ir amžiaus grupėse. Informacinių šaltinių ir priemonės, padedančios atpažinti retą ligą. Indikacijos genetiko konsultacijai. Indikacijos prenatalinei diagnostikai. Nukreipimas specializuotai diagnostikai. Genetinė diagnostika: citogenetiniai, biocheminiai genetiniai, molekuliniai genetiniai tyrimai. Naujos kartos sekoskaitos tyrimai. Integravotų omikos tyrimų svarba retų ligų diagnostikai. Bendradarbiavimo platformos ir dalijimasis duomenimis. Kiti diagnostiniai tyrimai retų ligų diagnostikoje: patologiniai, instrumentiniai, laboratoriniai.

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Specializuotas retų ligų gydymas

Paskaita. 2 val. Retieji vaistiniai preparatai: nuo klinikinių tyrimų iki ligonio lovos. Retų ligų vaistinių preparatų prieinamumas. Pažangioji terapija: genų, laštelių terapija. Farmakogenetika ir farmakogenomika. Personalizuota medicina. Sudėtingos chirurginės intervencijos ir transplantacija. Specializuoto gydymo kokybės ir saugumo užtikrinimas. Daugiadisciplinės komandos organizavimas. Retų ligų kompetencijos ir referencijos centralai. Europos referencijos centrų tinklai. E-sveikatos sprendimai.

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Integruota sergančiųjų retomis ligomis sveikatos priežiūra

Paskaita. 1 val. Daugiadisciplinės integruotos priežiūros svarba ir principai. Pacientų kelių organizavimas. Pirminės sveikatos priežiūros grandies vaidmuo pacientų keliuose. Tarpsektorinis bendradarbiavimas: sveikatos priežiūros, reabilitacijos, psichologinės, bendruomeninės, edukacinės, kitos paslaugos. Paslaugų koordinavimas ir atvejo vadyba. Pereinamųjų laikotarpių valdymas. Specifiniai integruotos priežiūros klausimai: paliatyvioji pagalba, perinatalinė priežiūra, ūmių būklių valdymas. E-sveikatos ir komunikacinių sprendimų.

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Genetinės ir retos ligos slaugytojo praktikoje

Paskaita. 2 val. Slaugytojo vaidmuo integruotoje sergančiųjų retomis ligomis priežiūroje. Išplėstinė slaugos. Priežiūros koordinavimas, atvejo vadyba, pereinamujų laikotarpių valdymas. Komandinio daugiadisciplinės priežiūros darbo organizavimas. Komunikacija su sergančiaisiais ir artimaisiais. Ligų prevencija, profilaktinės ir tikrinimo programos, rizikos vertinimas. Paciento ir artimujų mokymas ir stiprinimas. Genetinių paslaugų organizavimas.

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Genetiko vaidmuo daugiaudisciplinėje retų ligų priežiūroje

Paskaita. 2 val. Genetinės konsultacijos struktūra ir principai. Dismorfologinė ir sindromologinė fenotipo analizė. Diagnostiniai genetiniai tyrimai. Nešiotojų tyrimai. Ikišimptominiai tyrimai. Populiacinės tikrinimo programos. Genetinio ištyrimo strategijos parinkimas. Genetinių variantų klasifikacija, neaiškios klinikinės reikšmės genetiniai variantai. Genetinės diagnostikos iššūkiai: mozaicizmas, naujai atsiradusios mutacijos, oligogeninis paveldėjimas ir modifikuojantys variantai, somatinės mutacijos. Pakartotinės genetinės konsultacijos organizavimas: neaiškios klinikinės reikšmės genetiniai variantai, nediagnozuota genetinė liga. Atsitiktiniai radiniai. Genealogijos braižymas, paveldėjimo būdo nustatymas ir rizikos vertinimas. Kaskadinis šeimų ištyrimas. Pomirtiniai genetiniai tyrimai. E-sveikatos sprendimai ir telegenetika.

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Retų ligų prevencija

Paskaita. 2 val. Prevencijos programų svarba ir organizavimas. Teratogeniniai veiksniai: biologiniai, cheminiai, fiziniai. Teratogeninės rizikos vertinimas, konsultavimas, priežiūra. Teratogeniniai sindromai: alkoholinio vaisiaus sindromo spektras, valproatų poveikio vaisiui sindromas. Naujaginių

tikrinimo programų organizavimas, iššūkiai ir sprendimai. Prenatalinė priežiūra: tikrinimas ir diagnostika.

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Paciento ir šeimos stiprinimas

Seminaras. 2 val. Sergančiųjų retomis ligomis patirtis ir žinios apie retąsias ligas, komunikacija su sveikatos priežiūros darbuotojais. Pacientų ir šeimų poreikiai. Psichologinė ir emocinė pagalba. Savarankiška sveikatos priežiūra: iššūkiai, paciento mokymas ir stiprinimas. Specialistų ir pacientų bei šeimų bendradarbiavimas sprendimų priėmimo procese. Savitarpio pagalbos grupės, pacientų organizacijos. Informacinės ir komunikacinės priemonės. Pacientų organizacijos. Visuomenės švietimas.

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Retų ligų moksliniai tyrimai

Paskaita. 2 val. Retų ligų mokslinių tyrimų iššūkiai. Mokslinių tyrimų infrastruktūra: biobankai, registratoriai, bendradarbiavimo platformos. Mažų populiacijų klinikiniai tyrimai ir alternatyvios indikacijos vaistiniams preparatams. Bendradarbiavimo svarba: Europos jungtinė programa „Retos ligos“ ir Tarptautinis retų ligų mokslinių tyrimų konsorciumas. Bendradarbiavimas su pacientais ir pacientų organizacijomis retų ligų moksliniuose tyrimuose. Pacientų pranešamos sveikatos baigtys retų ligų moksliniuose tyrimuose. Bioetiniai ir teisiniai retų ligų mokslinių tyrimų klausimai. Intelektinė nuosavybė ir patentavimas. Duomenų valdymas pagal FAIR principus, apsauga, dalijimasis duomenimis. Dirbtinio intelekto panaudojimas retų ligų moksliniuose tyrimuose.

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Bioetiniai ir teisiniai klausimai

Seminaras. 2 val. Informuotas sutikimas. Duomenų apsauga ir privatumas. Bioetiniai ir teisiniai klausimai klinikinės genetikos ir personalizuotos medicinos praktikoje. Paslaugų prieinamumas. Pažeidžiamos populiacijos: vaikai, neįgalūs asmenys. Tiesiogiai vartotojui atliekami tyrimai. Genomo redagavimas. Genetinė diskriminacija ir eugenika.

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Daugiadisciplinė sergančiųjų paveldimomis medžiagų apykaitos ligomis sveikatos priežiūra

Seminaras. I val. Paveldimų medžiagų apykaitos ligų klasifikacija. Ūmiomis būklėmis pasireiškiančios paveldimos medžiagų apykaitos ligos: pirminė diagnostika, nukreipimas specializuotoms paslaugoms, ilgalaikis ūmių būklių valdymas. Organų ir organizmo sistemų pažeidimas, sergant paveldimomis medžiagų apykaitos ligomis. Specializuotas paveldimų medžiagų apykaitos ligų gydymas: mitybinis ir farmakologinis gydymas, pakaitinė fermentų terapija, genų ir ląstelių terapija. Daugiadisciplinės, integruotos priežiūros organizavimas. Specifiniai integruotos priežiūros klausimai: pereinamieji laikotarpiai, perinatalinė priežiūra, paliatyvioji pagalba, suaugusiuju priežiūra. Paveldimų medžiagų apykaitos ligų Europos referencijos centrų tinklas MetabERN.

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Daugiadisciplinė sergančiųjų retomis neurologinėmis ligomis sveikatos priežiūra

Seminaras. 1 val. Neurogenetinių ligų diagnostika. Reti genetiniai ekstrapiramidiniai judesių sutrikimai: distonija, ataksija, choréja. Smegenelių ataksijos. Hantingtono liga. Vilsono liga. Spastinė paraplegija. Daugiadisciplinės integruotos priežiūros organizavimas sergantiems retomis neurologinėmis ligomis. Retų neurologinių ligų Europos referencijos centrų tinklas ERN RND.

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Daugiadisciplinė sergančiųjų retomis nervų ir raumenų ligomis sveikatos priežiūra

Seminaras. 1 val. Retos nervų ir raumenų ligos: miopatijos, raumenų distrofijos, miotoninės ligos, spinalinė raumenų atrofija, polineuropatijos. Genetinis ištyrimas. Distrofinopatijos. Miotoninė distrofija. Raumenų kanalopatijos. Spinalinė raumenų atrofija. Paveldimos sensomotorinės polineuropatijos. Pažangi retų nervų ir raumenų ligų terapija. Daugiadisciplinės, integruotos priežiūros organizavimas sergantiems retomis nervų ir raumenų ligomis. Retų nervų ir raumenų Europos referencijos centrų tinklas Euro NMD.

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Daugiadisciplinė sergančiųjų retomis epilepsijomis sveikatos priežiūra

Seminaras. 1 val. Retų genetinių epilepsijų etiologinė struktūra. Vystymosi ir epileptinės encefalopatijos. Žievės malformacijos. Paveldimos medžiagų apykaitos ligos, pasireiškiančios epilepsija ar traukuliais. Genetiniai sindromai, pasireiškiantys epilepsija. Genetinės diagnostikos principai ir iššūkiai. Specializuotas retų epilepsijų gydymas: mitybinis ir farmakologinis gydymas, genų ir ląstelių terapija. Daugiadisciplinės, integruotos priežiūros organizavimas sergantiesiems retomis epilepsijomis. Retų epilepsijų Europos referencijos centrų tinklas EpiCARE.

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Daugiadisciplinė pacientų su retais klausos sutrikimais sveikatos priežiūra

Seminaras. 1 val. Klausos tikrinimo programos. Genetinės nesindrominio klausos sutrikimo priežastys. Genetiniai sindromai, pasireiškiantys klausos sutrikimu. Genetinė diagnostika. Daugiadisciplinė, integruota pacientų su klausos sutrikimais priežiūra.

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Daugiadisciplinė sergančiųjų retomis akių ligomis sveikatos priežiūra

Seminaras. 1 val. Retos akių ligos: retinopatijos, priekinio akies segmento sutrikimai, akių raidos ydos, akių judesių sutrikimai. Retinopatių oftalmologinė ir genetinė diagnostika. Genetinės kataraktos ir glaukomos priežastys. Genetiniai sindromai, pasireiškiantys užpakalinio ir priekinio akies segmento raidos ydomis ir pažeidimais. Pažangioji genetinių akių ligų terapija.

Daugiadisciplinė, integruota sergančiųjų retomis akių ligomis priežiūra. Retų akių ligų Europos referencijos centrų tinklas ERN EYE.

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Daugiadisciplinė pacientų su įgimtomis kraujagyslių anomalijomis sveikatos priežiūra

Seminaras. 1 val. Įgimtų kraujagyslių anomalijų klasifikacija. Pirminė įgimtų kraujagyslių anomalijų diagnostika ir nukreipimas specializuotoms paslaugoms. Specializuota įgimtų kraujagyslių anomalijų diagnostika. Specializuotas įgimtų kraujagyslių anomalijų gydymas: chirurginis, konservatyvus, farmakologinis. Genetinė diagnostika: monogeninės ligos ir somatinės mutacijos. Pagreitėjusio augimo genetiniai sindromai, pasireiškiantys įgimtomis kraujagyslių anomalijomis. Paveldimų hemoraginių telangiectazijų sindromas. Daugiadisciplinė, integruota pacientų su įgimtomis kraujagyslių anomalijomis priežiūra. Retų įgimtų kraujagyslių anomalijų Europos referencijos centrų tinklas VASCERN.

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Daugiadisciplinė sergančiųjų retomis endokrininės sistemos ligomis sveikatos priežiūra

Seminaras. I val. Retos genetinės endokrininės ligos, pažeidžiančios antinksčius, skydliaukę, kasos endokrininę dalį, sutrikdančios augimą ar lytinį brendimą. Igimta antinksčių hiperplazija. Igimta hipotirozė. Monogeninis diabetas. Augimo atsilikimas ir genetiniai sindromai, pasireiškiantys žemaūgiškumu. Lytinio brendimo sutrikimas ir genetiniai sindromai, pasireiškiantys brendimo sutrikimu. Prader Willi sindromas. Genetinė retų endokrininių ligų diagnostika. Daugadiplinė, integruota sergančiųjų retomis endokrininėmis ligomis priežiūra. Retų endokrininių ligų Europos referencijos centrų tinklas Endo ERN.

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Daugiadisciplinė pacientų su raidos anomalijomis ir intelektine negalia sveikatos priežiūra Seminaras. 1 val. Intelektinės negalios etiologinė struktūra. Autizmo spektro sutrikimų genetika. Dauginių raidos ydų genetika. Pirminė diagnostika raidos sutrikimo, intelektinės negalios ir raidos ydų atvejais: dismorfoliginis ir antropometrinis paciento ištyrimas. Žmogaus fenotipo ontologija, skaitmeninių įrankių panaudojimas fenotipo vertinimui. Dauno sindromas. Trapios X chromosomos sindromas. Genetinio ištyrimo strategija įgimtų raidos ydų ir intelektinės negalios atvejais. Daugiadisciplinė, integruota pacientų su įgimtomis raidos ydomis ir psichomotorinės raidos atsilikimu ar intelektine negalia priežiūra. Specializuotas raidos ydų gydymas, gydymo kokybės ir saugumo

užtikrinimas. Retų raidos anomalijų ir intelektinės negalios Europos referencijos centrų tinklas ERN ITHACA. Retų įgimtų raidos ydų Europos referencijos centrų tinklas ERNICA.

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Daugiadisciplinė sergančiųjų retomis širdies ligomis sveikatos priežiūra

Seminaras. 1 val. Retos širdies ligos: kardiomiopatijos, įgimti širdies ritmo ir laidumo sutrikimai, paveldimi lipidų apykaitos sutrikimai, pasireiškiantys širdies pažeidimu. Kardiomiopatių genetika: hipertrofinė, diliatacinė, aritmogeninė dešiniojo skilvelio, nekompaktinio miokardo kardiomiopatijos. Ilgo QT sindromai. Šeiminė hipercholesterolemija. Retų širdies ligų genetinė diagnostika: galimybės ir iššūkiai, kaskadinis šeimų ištyrimas. Genetika prevencinėje kardiologijoje. Įgimtų širdies ydų genetika. Daugiadisciplinė, integruota sergančiųjų retomis širdies ligomis priežiūra. Retų širdies ligų Europos referencijos centrų tinklas ERN GUARD HEART.

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Daugiadisciplinė sergančiųjų retomis inkstų ligomis sveikatos priežiūra

Seminaras. 1 val. Retos genetinės inkstų ligos: glomerulopatijos, ciliopatijos, tubulopatijos, metablinės nefropatijos. Igimtas nefrotinis sindromas. Policistinė inkstų liga. Alporto sindromas. Genetinės inkstų akmenligės formos, cistinurija. Cistinozė. Urogenitalinės raidos ydos. Genetinis ištyrimas. Specializuotas gydymas. Daugiadisciplinė, integruota sergančiųjų retomis inkstų ligomis priežiūra. Retų inkstų ligų Europos referencijos centrų tinklas ERKNet.

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Daugiadisciplinė sergančiųjų retomis plaučių ligomis sveikatos priežiūra

Seminaras. 1 val. Retų plaučių ligų etiologinė struktūra. Cistinė fibrozė. Plautinės arterijos hipertenzija. α 1-antitripsino stoka. Pirminė retų plaučių ligų diagnostika ir nukreipimas specializuotoms paslaugoms. Genetinė retų plaučių ligų diagnostika. Specializuotas gydymas. Daugiadisciplinė, integruota sergančiųjų retomis plaučių ligomis priežiūra. Retų plaučių ligų Europos referencijos centrų tinklas ERN LUNG.

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Daugiadisciplinė sergančiųjų retomis odos ligomis sveikatos priežiūra

Seminaras. 1 val. Retų genetinių odos ligų etiologinė struktūra. Pūslinė epidermolizė. Paveldima ichtiozė. Ektoderminė displazija. Mozaikinės odos ligos. Genetiniai sindromai, pasireiškiantys odos pažeidimu. Genetinė diagnostika. Daugiadisciplinė, integruota sergančiųjų retomis odos ligomis priežiūra. Retų odos ligų Europos referencijos centrų tinklas ERN Skin.

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Daugiadisciplinė sergančiųjų retomis skeleto ir jungiamojo audinio ligomis sveikatos priežiūra Seminaras. 1 val. Retų genetinių kaulų ligų etiologinė struktūra. Skeleto displazijos. Dauginių osteochondromų sindromas. Nebaigtinė osteogenėzė. Osteopetrozės. Hipofosfatazija. Marfano sindromas. Ehlers-Danlos sindromas. Paveldimos aortopatijos. Pirminė diagnostika ir nukreipimas specializuotoms paslaugoms. Genetinė diagnostika. Specializuotas gydymas: chirurginės intervencijos, pakaitinė fermentų terapija. Daugiadisciplinė, integruota sergančiųjų retomis skeleto ir jungiamojo audinio ligomis priežiūra. Retų kaulų ligų Europos referencijos centrų tinklas ERN BOND.

Rekomenduojama literatūra.

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