# Supplementary Materials

## Knowledge, Attitudes and Practices Regarding Prenatal Screening and Genetic Diagnosis of Chromosomal Anomalies Among Health Workers in Turkistan Region, Kazakhstan: A Cross-Sectional Survey of a Predominantly Primary-Care Workforce

Zhansaya Torgauytova, Ardak Ayazbekov, Sundetgali Kalmakhanov, Almagul Kurmanova, Seçil Özkan, Saken Khaidarov, Gaukhar Kurmanova, Arman Nurmakhanbetov, Aknur Turgumbayeva, Amina Abdrakhmanova, Diana Sundetova, Shynar Zakaryanova and Elnur Adilova

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### Contents

• Supplementary Material S1. Questionnaire (Kazakh original and English translation) with item codes, response distributions and scoring key
• Supplementary Material S2. Ethics conclusion and KazREC certificate (separate file, unchanged)
• Table S1. Multiple-response questions, overall and by level of care
• Table S2. CHERRIES checklist
• Table S3. Knowledge items by level of care
• Table S4. Item responses by level of care
• Table S5. Former attitude composite
• Table S6. Selected exploratory cross-tabulations
• Figure S1. Location of Turkistan region relative to the national medical genetics services
• Figure S2. Directed acyclic graph for the practice models

**Note on denominators.** Item-level tallies in S1 and Tables S1, S3 (by level of care), S4 and S6 come from the institutional cross-tabulation export, which holds 1330 records (1150 primary care, 180 hospital or perinatal unit). The analysis file used in the manuscript holds 1331 records (1146 primary care, 95 hospital, and 90 perinatal unit). The two files were exported separately and differ by one record and in how the five records are grouped by workplace. Percentages here use the export denominators; the manuscript uses 1331.

# Supplementary Material S1. Questionnaire and scoring key

## S1.1. Structure and administration

The questionnaire was written by the study team in Kazakh and administered online through Google Forms between 1 June and 30 July 2026. It has 38 items: 35 content items in four blocks, reproduced in full below, and three respondent items (consent, profession and workplace; S1.2). By domain, the content items comprise 14 knowledge items (11 single-choice and 3 multiple-response), 11 attitude and perception items, and 10 practice items. Five content questions allowed several answers: regulatory documents (KD1), diagnostic methods (KD2), components of combined screening (K11), barriers (BR), and reasons for not referring for invasive diagnosis (RR). All items were mandatory. Free-text answers were possible for some items; they are listed as "free text" below. The English translation was prepared for publication by Z.T. and back-translated by an independent bilingual public health specialist.

## S1.2. Respondent items

| Item | Question | Response options | n (%) |
|---|---|---|---|
| C1 | Consent to take part | Yes (required to proceed) | 1331 (100) |
| R1 | Profession | Nurse / midwife / general practitioner / obstetrician-gynecologist / other specialist (neonatologist, geneticist, ultrasound specialist) | 902 / 196 / 159 / 59 / 15 |
| R2 | Workplace | Primary care (polyclinic, primary health care centre, feldsher-midwife post) / hospital / perinatal care unit | 1146 / 95 / 90 |

Counts from the analysis file (n = 1331).

## S1.3. Content items with response distribution and scoring key

For each item the Kazakh original is followed by the English translation. Counts are from the institutional export (n = 1330). Score column: 1/0, knowledge key; 2/1/0, ordinal coding (practice items are summed into the 7-item practice score; attitude items are reported individually and no longer summed); ✓/✗, options required/excluded for a correct answer on a multiple-response item; R/D, relevant document/distractor; —, not scored.

### Block 1. Awareness and knowledge of prenatal screening

| Code | Kazakh original | English translation | n (%) | Score |
|---|---|---|---|---|
| **AW · Perception · single choice** | | | | |
| | Өзіңіз қызмет атқаратын өңірде медико-генетикалық көмек көрсету қызметінің бар екенін білесіз бе? | Do you know whether a medical genetics service operates in the region where you work? | | |
| | Иә, толық | Yes, fully | 890 (66.9) | 2 |
| | Ішінара | Partly | 267 (20.1) | 1 |
| | Хабарым жоқ | Not aware | 173 (13.0) | 0 |
| | | Note: Covariate (awareness, 0–2); not part of any score. | | |
| **KD1 · Knowledge (descriptive) · multiple response** | | | | |
| | Қазақстан Республикасында жүктілік кезіндегі пренаталды скринингті реттейтін негізгі нормативтік құжаттар қандай? | Which are the main regulatory documents governing prenatal screening during pregnancy in the Republic of Kazakhstan? | | |
| | ҚР ДСМ № 272 бұйрығы, 27 сәуір 2015 ж. «Денсаулық сақтау саласындағы мемлекеттік көрсетілетін қызметтер стандарттарын бекіту туралы» | Order No. 272 of 27 April 2015 on state service standards in health care (distractor) | 81 (6.1) | D |
| | ҚР ДСМ № 52 бұйрығы, 31 наурыз 2023 – неонатальды көмек көрсету стандарттары | Order No. 52 of 31 March 2023, standards of neonatal care (distractor) | 169 (12.7) | D |
| | ҚР ДСМ № ҚР ДСМ-54 (2022) «Стандартты пренаталды ультрадыбыстық скрининг жүргізу | Order No. KR DSM-54 (2022), standard for organizing prenatal ultrasound screening | 397 (29.8) | R |

| Code | Kazakh original | English translation | n (%) | Score |
|---|---|---|---|---|
| | ұйымының стандарты» | | | |
| | ҚР ДСМ № 704 бұйрығы(2010) «Скринингті ұйымдастыру қағидаларын бекіту туралы» | Order No. 704 (2010), rules for organizing screening | 406 (30.5) | R |
| | ҚР ДСМ № 92 бұйрығы (2021 жылғы 26 тамыз) «Қазақстан Республикасында акушерлік-гинекологиялық көмекті ұйымдастыру стандарты туралы» | Order No. ҚR DSM-92 (26 August 2021), standard for organizing obstetric and gynecological care | 934 (70.2) | R |
| | | Note: Descriptive only. R, relevant document; D, distractor. | | |
| **KD2 · Knowledge (descriptive) · multiple response** | | | | |
| | Сіз жүктілік кезінде жасалатын қандай генетикалық диагностика әдістерін білесіз? | Which genetic diagnostic methods used during pregnancy do you know? | | |
| | Пренатальды ультрадыбыстық (УДЗ) скрининг | Prenatal ultrasound screening | 1027 (77.2) | ✓ |
| | Биохимиялық скрининг (PAPP-A, β-ХГЧ және т.б.) | Biochemical screening (PAPP-A, β-hCG, etc.) | 813 (61.1) | ✓ |
| | Инвазивті емес пренатальды тестілеу (НИПТ) | Non-invasive prenatal testing (NIPT) | 147 (11.1) | ✓ |
| | Инвазивті диагностика (амниоцентез, хорион биопсиясы) | Invasive diagnosis (amniocentesis, chorionic villus sampling) | 295 (22.2) | ✓ |
| | Молекулалық-генетикалық тесттер | Molecular genetic tests | 364 (27.4) | ✓ |
| | Білмеймін | I do not know | 30 (2.3) | — |
| | | Note: Descriptive only (number of methods recognized, 0–5). | | |
| **K1 · Knowledge · single choice** | | | | |
| | Пренаталды генетикалық скринингтің негізгі мақсаттары қандай? | What are the main aims of prenatal genetic screening? | | |
| | Хромосомалық ақаулардың даму қаупін анықтау | Assessing the risk of chromosomal anomalies | 1223 (92.0) | 1 |
| | Нәрестенің жынысын анықтау | Determining fetal sex | 55 (4.1) | 0 |
| | Плацента қызметін бағалау | Assessing placental function | 52 (3.9) | 0 |
| **K8 · Knowledge · single choice** | | | | |
| | Жүктілік кезінде бірінші пренаталды ультрадыбыстық скрининг жүктіліктің қай аптасында жасалады? | In which week of pregnancy is the first prenatal ultrasound screening performed? | | |
| | 9+0-12+6 аптада | $9^{+0}$–$12^{+6}$ weeks | 350 (26.3) | 0 |
| | 10+0-14+6 аптада | $10^{+0}$–$14^{+6}$ weeks | 137 (10.3) | 0 |
| | 11+0-13+6 аптада | $11^{+0}$–$13^{+6}$ weeks | 651 (48.9) | 1 |
| | 12+0-14+6 аптада | $12^{+0}$–$14^{+6}$ weeks | 192 (14.4) | 0 |
| | | Note: Keyed to Order No. ҚR DSM-54. | | |
| **K10 · Knowledge · single choice** | | | | |
| | Жүктілік кезінде екінші пренаталды ультрадыбыстық скрининг жүктіліктің қай аптасында жасалады? | In which week of pregnancy is the second prenatal ultrasound screening performed? | | |
| | 16+0-20+0 аптада | $16^{+0}$–$20^{+0}$ weeks | 215 (16.2) | 0 |
| | 18+0-21+6 аптада | $18^{+0}$–$21^{+6}$ weeks | 401 (30.2) | 0 (1 in sensitivity analysis) |
| | 19+0-21+0 аптада | $19^{+0}$–$21^{+0}$ weeks | 483 (36.3) | 1 |
| | 19+6-21+0 аптада | $19^{+6}$–$21^{+0}$ weeks | 231 (17.4) | 0 |
| | | Note: Keyed to Order No. ҚR DSM-54; $18^{+0}$–$21^{+6}$ | | |

| Code | Kazakh original | English translation | n (%) | Score |
|---|---|---|---|---|
| | | weeks also accepted in a sensitivity analysis. | | |
| **K4 · Knowledge · single choice** | | | | |
| | Жүктілік кезіндегі даму ақаулары және хромосомдық ауытқуларды анықтауда1-ші триместірінде жасалатын ана қаны сарысуының (МСМ) биохимиялық маркерлерінің қай түрлері ҚР қолданылады | Which first-trimester maternal serum biochemical markers are used in Kazakhstan to detect developmental defects and chromosomal anomalies? | | |
| | РАРР-А, β-ХГЧ | PAPP-A, β-hCG | 776 (58.3) | 1 |
| | РАРР-А, β-ХГЧ, АФП | PAPP-A, β-hCG, AFP | 477 (35.9) | 0 |
| | Ингибин А | Inhibin A | 26 (2.0) | 0 |
| | Конъюгирленбеген эстрадиол | Unconjugated estradiol | 51 (3.8) | 0 |
| **K11 · Knowledge · multiple response** | | | | |
| | Жүктіліктің бірінші триместрінде жүргізілетін комбинирленген пренаталды скрининг қандай зерттеулерден тұрады? | Which examinations make up combined prenatal screening in the first trimester? | | |
| | Ультрадыбыстық зерттеу | Ultrasound examination | 1042 (78.3) | ✓ |
| | Биохимиялық маркерлерді анықтау (РАРР-А, β-ХГЧ) | Biochemical markers (PAPP-A, β-hCG) | 930 (69.9) | ✓ |
| | Инвазивті тексеру (амниоцентез) | Invasive testing (amniocentesis) | 153 (11.5) | ✗ |
| | Конъюгирленбеген эстриол | Unconjugated estriol | 22 (1.7) | ✗ |
| | АФП және ингибин А | AFP and inhibin A | 50 (3.8) | ✗ |
| | Жүктілікті визуалды бақылау | Visual follow-up of the pregnancy | 189 (14.2) | ✗ |
| | | Note: Multiple response. Scored 1 only if both ✓ options and no ✗ option were selected (n = 483, 36.3%). | | |
| **K3 · Knowledge · single choice** | | | | |
| | Кариотиптеу дегеніміз не? | What is karyotyping? | | |
| | Хромосомалар саны мен құрылымын зерттеу әдісі | A method for studying chromosome number and structure | 835 (62.8) | 1 |
| | Геннің мутациясын анықтау әдісі | A method for detecting gene mutations | 243 (18.3) | 0 |
| | УДЗ арқылы ұрықтың даму кезеңін бағалау | Assessing fetal developmental stage by ultrasound | 191 (14.4) | 0 |
| | Ағзадағы гормон деңгейін бағалау | Assessing hormone levels in the body | 61 (4.6) | 0 |
| **K9 · Knowledge · single choice** | | | | |
| | Жүкті әйелдер қай жастан туа біткен және тұқым қуалайтын патологияның дамуы бойынша жоғары қаупі тобына жатады? | From what age are pregnant women considered at high risk of congenital and hereditary pathology? | | |
| | 30 жастан бастап | From 30 years | 181 (13.6) | — |
| | 32 жастан бастап | From 32 years | 36 (2.7) | — |
| | 35 жастан бастап | From 35 years | 550 (41.4) | 1 (analysis key) |
| | 37 жастан бастап | From 37 years | 148 (11.1) | National orders |
| | 40 жастан бастап | From 40 years | 415 (31.2) | — |
| | | Note: Keyed to 35 years in the analysis; Orders No. 704 and No. ҚR DSM-92 use 37 years, and in combined screening age is one input into individual risk (see S1.5). | | |
| **K12 · Knowledge · single choice** | | | | |
| | Жүктіліктің1-ші триместіріндегі пренаталдық | In what unit are the results of first-trimester | | |

| Code | Kazakh original | English translation | n (%) | Score |
|---|---|---|---|---|
| | (комбинирленген) скрининг нәтижелері қандай бірлікпен интерпретацияланады? | (combined) prenatal screening interpreted? | | |
| | МоМ | MoM (multiple of the median) | 218 (16.4) | 1 |
| | МЕ/л | IU/L | 499 (37.5) | 0 |
| | г/л | g/L | 290 (21.8) | 0 |
| | ММ | "MM" (abbreviation as printed on the form) | 323 (24.3) | 0 |

## Block 2. Attitudes and perceptions

| Code | Kazakh original | English translation | n (%) | Score |
|---|---|---|---|---|
| **A2 · Attitude · single choice** | | | | |
| | Сіз барлық жүкті әйелдерге ұрықтың даму ақауы және хромосомдық аномалияны анықтау үшін пренатальды скринингті міндетті түрде жүргізу қажет деп ойлайсыз ба? | Do you think prenatal screening for fetal malformations and chromosomal anomalies should be mandatory for all pregnant women? | | |
| | Иә, міндетті түрде | Yes, mandatory | 1126 (84.7) | 2 |
| | Иә, ұсынылады | Yes, recommended | 109 (8.2) | 1 |
| | Тек көрсеткіштер бойынша | Only when indicated | 66 (5.0) | 1 |
| | Жоқ | No | 29 (2.2) | 0 |
| | | Note: Reported individually; former composite coding shown. | | |
| **A1 · Attitude · single choice** | | | | |
| | Туа біткен және тұқым қуалайтын патология бойынша «қауіп тобына» жататын жүкті әйелдерді генетик дәрігерге жолдауға көзқарасыңыз | What is your attitude to referring pregnant women at risk of congenital and hereditary pathology to a geneticist? | | |
| | Міндетті деп есептеймін | I consider it mandatory | 1161 (87.3) | 2 |
| | Ұсынылады, бірақ міндетті емес | Recommended but not mandatory | 106 (8.0) | 1 |
| | Қажеті жоқ, ерікті түрде болу керек | Not needed, should be voluntary | 38 (2.9) | 1 |
| | Теріс қараймын | Negative | 25 (1.9) | 0 |
| | | Note: Reported individually; former composite coding shown. | | |
| **TOP · Perception · single choice** | | | | |
| | Сіздің пікіріңізше, ауыр емдеуге келмейтін тұқым қуалайтын аурулар анықталған жағдайда медициналық көрсеткіштер бойынша жүктілікті үзу дұрыс па? | In your opinion, is termination of pregnancy on medical grounds appropriate when a severe untreatable hereditary disease is detected? | | |
| | Иә | Yes | 1029 (77.4) | — |
| | Жауап беруге қиналамын | Difficult to answer | 253 (19.0) | — |
| | Жоқ | No | 48 (3.6) | — |
| | | Note: Reported descriptively; never scored (ethical position). | | |
| **PAW · Perception · single choice** | | | | |
| | Халықтың медико-генетикалық қызмет мүмкіндіктері туралы ақпараттану деңгейін қалай бағалайсыз? | How do you rate the population's awareness of what medical genetics services offer? | | |
| | Жоғары | High | 597 (44.9) | — |
| | Орташа | Moderate | 634 (47.7) | — |
| | Төмен | Low | 99 (7.4) | — |

| Code | Kazakh original | English translation | n (%) | Score |
|---|---|---|---|---|
| | | Note: Descriptive only. | | |
| **AC · Perception · single choice** | | | | |
| | Сіз өз өңіріңізде жүкті әйелдерге медико-генетикалық көмектің қолжетімділігін қалай бағалайсыз? (1-ден 5-ке дейінгі шкала бойынша бағалаңыз) | How do you rate the accessibility of medical genetic care for pregnant women in your region? (scale 1–5) | | |
| | 1 – Қол жетімді емес | 1 – Not accessible | 149 (11.2) | 1 |
| | 2 – Қиындықпен қолжетімді | 2 – Accessible with difficulty | 110 (8.3) | 2 |
| | 3 – Ішінара қолжетімді | 3 – Partly accessible | 207 (15.6) | 3 |
| | 4 – Жалпы қолжетімді | 4 – Generally accessible | 496 (37.3) | 4 |
| | 5 – Толығымен қолжетімді | 5 – Fully accessible | 368 (27.7) | 5 |
| | | Note: Covariate (1–5). | | |
| **BR · Perception · multiple response** | | | | |
| | Жүкті әйелдердің генетикалық көмек алуындағы негізгі кедергілер қандай? | What are the main barriers to pregnant women receiving genetic care? | | |
| | Мамандардың жоқтығы | Shortage of specialists | 567 (42.6) | — |
| | Жүкті әйелдердің жеткіліксіз ақпараттандырылуы | Inadequate information for pregnant women | 508 (38.2) | — |
| | Географиялық алшақтық | Geographical distance | 361 (27.1) | — |
| | Қаржылық шектеулер (қызметтер тек ақылы) | Financial constraints (services are fee-based only) | 294 (22.1) | — |
| | Медицина қызметкерлерінің жеткіліксіз хабардар болуы | Insufficient awareness among health workers | 160 (12.0) | — |
| | | Note: Multiple response; descriptive. | | |
| **A5 · Attitude · single choice** | | | | |
| | Сіздің пікіріңізше көрсеткіш болған жағдайда инвазивті пренаталды диагностика жасау қажет пе? | In your opinion, is invasive prenatal diagnosis necessary when indicated? | | |
| | Қажет, міндетті түрде | Necessary, mandatory | 1230 (92.5) | 2 |
| | Жүктінің келісуімен | With the pregnant woman's consent (free text) | 3 (0.2) | — |
| | Қажеті жоқ | Not necessary | 91 (6.8) | 0 |
| | Билмеймин | I do not know (free text) | 6 (0.5) | 0 |
| | | Note: Reported individually (main text, Table 4). | | |
| **A4 · Attitude · single choice** | | | | |
| | Медициналық генетика саласындағы қызметтерді дамыту еліміз үшін маңызды деп ойлайсыз ба? | Do you think developing medical genetics services is important for the country? | | |
| | Иә, өте маңызды | Yes, very important | 973 (73.2) | 2 |
| | Иә, маңызды | Yes, important | 298 (22.4) | 1 |
| | Жауап беру қиын | Difficult to answer | 46 (3.5) | 0 |
| | Жоқ, маңызды емес | No, not important | 13 (1.0) | 0 |
| | | Note: Reported individually; former composite coding shown. | | |
| **A6 · Self-assessment · single choice** | | | | |
| | Жүктілік кезіндегі ұрықтың даму ақаулары және хромосмдық аномалияларын анықтау барысындағы пренатальды генетикалық | How confident are you in your knowledge of prenatal genetic screening for fetal malformations and chromosomal anomalies? | | |

| Code | Kazakh original | English translation | n (%) | Score |
|---|---|---|---|---|
| | скрининг бойынша өз біліміңізге қаншалықты сенімдісіз? | | | |
| | Толығымен сенімдімін | Fully confident | 517 (38.9) | 2 |
| | Негізгі аспектілерін білемін | I know the main aspects | 251 (18.9) | 1 |
| | Қосымша білім қажет | I need further training | 516 (38.8) | 1 |
| | Тақырыпты түсінбеймін | I do not understand the topic | 46 (3.5) | 0 |
| | | Note: Self-assessment; reported separately. | | |
| **A3 · Attitude · single choice** | | | | |
| | Генетикалық кеңес берудің енгізілуі тұқым қуалайтын аурулары бар балалардың туылу жиілігін төмендетеді деп ойлайсыз ба? | Do you think introducing genetic counselling reduces the birth rate of children with hereditary diseases? | | |
| | Иә | Yes | 1169 (87.9) | 2 |
| | Мумкін | Maybe (free text) | 8 (0.6) | 1 |
| | Жоқ | No | 152 (11.4) | 0 |
| | Түсінбедім | Did not understand (free text) | 1 (0.1) | 0 |
| | | Note: Offered options were yes and no; other answers are free text. | | |

## Block 3. Clinical and counselling practice

| Code | Kazakh original | English translation | n (%) | Score |
|---|---|---|---|---|
| **P7 · Practice · single choice** | | | | |
| | Соңғы бір жыл ішінде пациенттерді медициналық генетик дәрігеріне консультацияға жолдадыңызба? | In the past year, have you referred patients to a medical geneticist for consultation? | | |
| | Жиі | Frequently | 569 (42.8) | 2 |
| | Кейде | Sometimes | 434 (32.6) | 1 |
| | Жолдамадым | Did not refer | 327 (24.6) | 0 |
| | | Note: Physician task under the national orders. | | |
| **P5 · Practice · single choice** | | | | |
| | Медико-генетикалық зерттеулердің нәтижелерін өз практикаңызда қолданасыз ба? | Do you use the results of medical genetic tests in your practice? | | |
| | Иә, тұрақты түрде | Yes, regularly | 641 (48.2) | 2 |
| | Кейде | Sometimes | 477 (35.9) | 1 |
| | Ешқашан | Never | 212 (15.9) | 0 |
| | | Note: Physician task under the national orders. | | |
| **P4 · Practice · single choice** | | | | |
| | Пренатальды және неонатальды скринингтің маңыздылығы туралы пациенттермен әңгіме өткізесіз бе? | Do you talk with patients about the importance of prenatal and neonatal screening? | | |
| | Жиі | Frequently | 736 (55.3) | 2 |
| | Кейде | Sometimes | 460 (34.6) | 1 |
| | Ешқашан | Never | 131 (9.8) | 0 |
| | Әрине | Of course (free text) | 3 (0.2) | 0* |
| | | Note: *Free-text answer coded 0 under the conservative rule. | | |
| **P1 · Practice · single choice** | | | | |

| Code | Kazakh original | English translation | n (%) | Score |
|---|---|---|---|---|
| | Пациенттерге (көрсеткіштер бойынша молекулярлы-генетикалық тестілеуден өтуге ұсыныс бересізбе? | Do you recommend molecular genetic testing to patients when indicated? | | |
| | Иә | Yes | 985 (74.1) | 2 |
| | Кейде | Sometimes | 245 (18.4) | 1 |
| | Жоқ | No | 100 (7.5) | 0 |
| | | Note: Physician task under the national orders. | | |
| **SRC · Practice (descriptive) · single choice** | | | | |
| | Жүктілік кезіндегі пренаталдық скрининг туралы ақпаратты қайдан аласыз? | Where do you get information about prenatal screening during pregnancy? | | |
| | Клиникалық нұсқаулықтар | Clinical guidelines | 667 (50.2) | EBM |
| | Ғылыми конференциялар / семинарлар | Scientific conferences or seminars | 188 (14.1) | EBM |
| | Әріптестерден | Colleagues | 253 (19.0) | — |
| | Интернет/әлеуметтік желілер | Internet or social media | 212 (15.9) | — |
| | Әрдайым | "Always" (free text) | 6 (0.5) | Other |
| | Кейде | "Sometimes" (free text) | 2 (0.2) | Other |
| | Поликлиника | "Polyclinic" (free text) | 1 (0.1) | Other |
| | Уч.мейірбикеден | "From the district nurse" (free text) | 1 (0.1) | Other |
| | | Note: Covariate: guidelines or conferences (EBM) versus other. | | |
| **P2 · Practice · single choice** | | | | |
| | Жүктілікті жоспарлап жүрген жұптарға тұқым қуалайтын аурулардың қаупі туралы әңгіме жүргізесіз бе? | Do you talk with couples planning a pregnancy about the risk of hereditary diseases? | | |
| | Иә, тұрақты түрде | Yes, regularly | 933 (70.2) | 2 |
| | Кейде | Sometimes | 309 (23.2) | 1 |
| | Жоқ | No | 88 (6.6) | 0 |
| **P6 · Practice · single choice** | | | | |
| | Пациенттердің молекулярлы-генетикалық тест нәтижелерін (мысалы, тұқым қуалайтын ауруларға күдіктенген жағдайда қаншалықты жиі аласыз? | How often do you receive patients' molecular genetic test results (e.g., when a hereditary disease is suspected)? | | |
| | Жиі | Frequently | 594 (44.7) | 2 |
| | Сирек | Rarely | 556 (41.8) | 1 |
| | Ешқашан | Never | 180 (13.5) | 0 |
| | | Note: Physician task under the national orders. | | |
| **P3 · Practice · single choice** | | | | |
| | Көрсеткіш болған жағдайда пациенттерге НИПТ (инвазивті емес пренатальды тестілеу) өтуге кеңес бересізбе? | Do you advise patients to undergo NIPT (non-invasive prenatal testing) when indicated? | | |
| | Иә, әрдайым | Yes, always | 844 (63.5) | 2 |
| | Кейде | Sometimes | 387 (29.1) | 1 |
| | Ешқашан | Never | 99 (7.4) | 0 |
| | | Note: Physician task under the national orders. | | |
| **TR · Exposure · single choice** | | | | |
| | Соңғы 3 жылда медициналық генетика бойынша оқу немесе семинарларға | Have you attended training or seminars on medical genetics in the last 3 years? | | |

| Code | Kazakh original | English translation | n (%) | Score |
|---|---|---|---|---|
| | қатыстыңыз ба? | | | |
| | Иә | Yes | 438 (32.9) | 1 |
| | Жоқ | No | 892 (67.1) | 0 |
| | | Note: Exposure; counted as 2/0 only in the eight-item sensitivity version of the practice index. | | |

## Block 4. Cytogenetics, screening stages and invasive diagnosis

| Code | Kazakh original | English translation | n (%) | Score |
|---|---|---|---|---|
| **K5 · Knowledge · single choice** | | | | |
| | Трисомиялар (Даун синдромы) қай хромосоманың аномалиясымен байланысты? | Trisomy (Down syndrome) is associated with an anomaly of which chromosome? | | |
| | 21-хромосома | Chromosome 21 | 772 (58.0) | 1 |
| | 13-хромосома | Chromosome 13 | 205 (15.4) | 0 |
| | 18-хромосома | Chromosome 18 | 81 (6.1) | 0 |
| | Х-хромосома | X chromosome | 269 (20.2) | 0 |
| | Жауап жоқ | No answer | 3 (0.2) | 0 |
| **K7 · Knowledge · single choice** | | | | |
| | Трисомиялар (Эдвардс синдромы) қай хромосомалардың аномалиясымен байланысты? | Trisomy (Edwards syndrome) is associated with an anomaly of which chromosome? | | |
| | 18-хромосома | Chromosome 18 | 686 (51.6) | 1 |
| | 13-хромосома | Chromosome 13 | 247 (18.6) | 0 |
| | 21-хромосома | Chromosome 21 | 199 (15.0) | 0 |
| | Х-хромосома | X chromosome | 194 (14.6) | 0 |
| | Жауап жоқ | No answer | 4 (0.3) | 0 |
| **K6 · Knowledge · single choice** | | | | |
| | Трисомиялар ( Патау синдромы) қай хромосомалардың аномалиясымен байланысты? | Trisomy (Patau syndrome) is associated with an anomaly of which chromosome? | | |
| | 13-хромосома | Chromosome 13 | 758 (57.0) | 1 |
| | 18-хромосома | Chromosome 18 | 205 (15.4) | 0 |
| | 21-хромосома | Chromosome 21 | 190 (14.3) | 0 |
| | Х-хромосома | X chromosome | 173 (13.0) | 0 |
| | Жауап жоқ | No answer | 4 (0.3) | 0 |
| **K2 · Knowledge · single choice** | | | | |
| | Жүктілік кезінде жасалатын пренаталды скринингтің қай кезеңдері ең маңызды? | Which stages of prenatal screening during pregnancy are the most important? | | |
| | 1-ші және 2-ші триместр | First and second trimester | 1194 (89.8) | 1 |
| | 3-ші триместр | Third trimester | 73 (5.5) | 0 |
| | Тек босану алдында | Only before delivery | 27 (2.0) | 0 |
| | Жүктілік кезінде скринингҚажет емес | Screening is not needed during pregnancy | 36 (2.7) | 0 |
| **RR · Practice (descriptive) · multiple response** | | | | |
| | Сіздің жүкті әйелдерді пренаталды инвазивті диагностика жасауға (мысалы, амниоцентез-қағанақ суын алып зерттеу) жолдаудан бас | What are your reasons for not referring pregnant women for invasive prenatal diagnosis (e.g., amniocentesis)? | | |

| Code | Kazakh original | English translation | n (%) | Score |
|---|---|---|---|---|
| | тартуыңыздың себебі? | | | |
| | Инвазивті әдістердің асқыну қаупінің болуы (жүктіліктің үзілуі, инфекция т.б.) | Risk of complications (pregnancy loss, infection, etc.) | 709 (53.3) | — |
| | Пациенттердің қорқынышы және инвазивті процедураны қабылдамауы | Patients' fear and refusal of the procedure | 634 (47.7) | — |
| | Медициналық этика немесе құқықтық мәселелер (рұқсат алу қиындығы) | Ethical or legal issues (difficulty obtaining permission) | 181 (13.6) | — |
| | Пациенттің әлеуметтік-экономикалық жағдайы (қаржылық мүмкіндіктері жоқ) | Patient's socioeconomic situation (cannot afford it) | 164 (12.3) | — |
| | Құрал-жабдықтардың немесе мамандардың жетіспеушілігі | Lack of equipment or specialists | 150 (11.3) | — |
| | Инвазивті диагностика нәтижелерін өңдеу мен түсіндіру қиындықтары | Difficulty processing and interpreting results | 146 (11.0) | — |
| | Медицина қызметкерлерінің тәжірибесінің жеткіліксіздігі ,білім деңгейінің төмендігі | Insufficient experience or knowledge of health workers | 108 (8.1) | — |
| | Басқа себептер | Other reasons | 134 (10.1) | — |
| | | Note: Multiple response; descriptive. | | |

## S1.4. Scoring rules

**Knowledge index (0–12).** One point was awarded for each correct answer on K1–K12. K11 (multiple response) scores 1 only when ultrasound and the PAPP-A/β-hCG option are both selected and no other option is selected. K9 (maternal age) was keyed to 35 years in the analysis; the national orders specify 37 years. The mean of the 11 items without K9, and with $18^{+0}$–$21^{+6}$ weeks also accepted for K10, is reported in the main text (Table 8).

**Practice index (0–14).** This is the sum of P1–P7 coded 2/1/0. Training attendance (TR) is analyzed as an exposure, not a practice; an eight-item version counting TR as 2/0 is used in a sensitivity analysis.

**Attitude items.** A1–A5 are reported individually; A6 (self-rated confidence) is a self-assessment and is reported separately. The previously summed attitude score (A1–A6, 0–12) is described in Table S5 and is used only as a covariate in the practice models.

**Categories.** For knowledge and practice: a score below 60% of the maximum, poor; 60–79.9% is moderate; 80% or above is good. These are operational cut-points, not validated competency standards.

**Free text.** Free-text answers that matched a predefined category were recoded to it; others were coded as 0 (practice) or "other" (information source).

## S1.5. Notes on the key

• K9: the analysis key (35 years) did not match Orders No. 704 and No. ҚР DSM-92 (37 years). This is an error in the answer key, not in data collection; the item and its options were presented as designed.
• K11: a multiple-response item, scored all-or-nothing as described in S1.4.
• K12: the question asked how results are interpreted.
• TR: this is not part of the practice index.
• Attitude items: these are not summed, because the coding ranked "mandatory" above "recommended", A6 measures confidence, and the sum had a ceiling effect.

## Table S1. Multiple-response questions, overall and by level of care

| Response | Overall, n (%) N = 1330 | 95% CI | Primary care, n (%) N = 1150 | Hospital or perinatal, n (%) N = 180 | p |
|---|---|---|---|---|---|
| **KD1. Which are the main regulatory documents governing prenatal screening during pregnancy in the Republic of Kazakhstan?** | | | | | |
| Order No. 272 of 27 April 2015 on state service standards in health care (distractor) | 81 (6.1) | 4.9–7.5 | 58 (5.0) | 23 (12.8) | <0.001 |
| Order No. 52 of 31 March 2023, standards of neonatal care (distractor) | 169 (12.7) | 11.0–14.6 | 149 (13.0) | 20 (11.1) | 0.549 |
| Order No. ҚР DSM-54 (2022), standard for organizing prenatal ultrasound screening | 397 (29.8) | 27.5–32.4 | 326 (28.3) | 71 (39.4) | 0.004 |
| Order No. 704 (2010), rules for organizing screening | 406 (30.5) | 28.1–33.1 | 317 (27.6) | 89 (49.4) | <0.001 |
| Order No. ҚР DSM-92 (26 August 2021), standard for organizing obstetric and gynecological care | 934 (70.2) | 67.7–72.6 | 820 (71.3) | 114 (63.3) | 0.035 |
| **KD2. Which genetic diagnostic methods used during pregnancy do you know?** | | | | | |
| Prenatal ultrasound screening | 1027 (77.2) | 74.9–79.4 | 874 (76.0) | 153 (85.0) | 0.007 |
| Biochemical screening (PAPP-A, β-hCG, etc.) | 813 (61.1) | 58.5–63.7 | 701 (61.0) | 112 (62.2) | 0.805 |
| Non-invasive prenatal testing (NIPT) | 147 (11.1) | 9.5–12.9 | 108 (9.4) | 39 (21.7) | <0.001 |
| Invasive diagnosis (amniocentesis, chorionic villus sampling) | 295 (22.2) | 20.0–24.5 | 228 (19.8) | 67 (37.2) | <0.001 |
| Molecular genetic tests | 364 (27.4) | 25.0–29.8 | 308 (26.8) | 56 (31.1) | 0.243 |
| I do not know | 30 (2.3) | 1.6–3.2 | 25 (2.2) | 5 (2.8) | 0.588 |
| **K11. Which examinations make up combined prenatal screening in the first trimester?** | | | | | |
| Ultrasound examination | 1042 (78.3) | 76.1–80.5 | 901 (78.3) | 141 (78.3) | >0.999 |
| Biochemical markers (PAPP-A, β-hCG) | 930 (69.9) | 67.4–72.3 | 802 (69.7) | 128 (71.1) | 0.793 |
| Invasive testing (amniocentesis) | 153 (11.5) | 9.9–13.3 | 127 (11.0) | 26 (14.4) | 0.208 |
| Unconjugated estriol | 22 (1.7) | 1.1–2.5 | 19 (1.7) | 3 (1.7) | >0.999 |
| AFP and inhibin A | 50 (3.8) | 2.9–4.9 | 43 (3.7) | 7 (3.9) | 0.835 |
| Visual follow-up of the pregnancy | 189 (14.2) | 12.4–16.2 | 165 (14.3) | 24 (13.3) | 0.818 |
| **BR. What are the main barriers to pregnant women receiving genetic care?** | | | | | |
| Shortage of specialists | 567 (42.6) | 40.0–45.3 | 506 (44.0) | 61 (33.9) | 0.012 |
| Inadequate information for pregnant women | 508 (38.2) | 35.6–40.8 | 405 (35.2) | 103 (57.2) | <0.001 |
| Geographical distance | 361 (27.1) | 24.8–29.6 | 317 (27.6) | 44 (24.4) | 0.418 |
| Financial constraints (services are fee-based only) | 294 (22.1) | 20.0–24.4 | 257 (22.3) | 37 (20.6) | 0.630 |
| Insufficient awareness among health workers | 160 (12.0) | 10.4–13.9 | 120 (10.4) | 40 (22.2) | <0.001 |
| **RR. What are your reasons for not referring pregnant women for invasive prenatal diagnosis (e.g., amniocentesis)?** | | | | | |
| Risk of complications (pregnancy loss, infection, etc.) | 709 (53.3) | 50.6–56.0 | 604 (52.5) | 105 (58.3) | 0.149 |
| Patients' fear and refusal of the procedure | 634 (47.7) | 45.0–50.4 | 547 (47.6) | 87 (48.3) | 0.873 |
| Ethical or legal issues (difficulty obtaining permission) | 181 (13.6) | 11.9–15.6 | 154 (13.4) | 27 (15.0) | 0.559 |
| Patient's socioeconomic situation (cannot afford it) | 164 (12.3) | 10.7–14.2 | 143 (12.4) | 21 (11.7) | 0.903 |
| Lack of equipment or specialists | 150 (11.3) | 9.7–13.1 | 130 (11.3) | 20 (11.1) | >0.999 |
| Difficulty processing and interpreting results | 146 (11.0) | 9.4–12.8 | 124 (10.8) | 22 (12.2) | 0.608 |
| Insufficient experience or knowledge of health workers | 108 (8.1) | 6.8–9.7 | 92 (8.0) | 16 (8.9) | 0.661 |
| Other reasons | 134 (10.1) | 8.6–11.8 | 116 (10.1) | 18 (10.0) | >0.999 |

Percentages sum to more than 100 because several answers were allowed. Wilson 95% CI. p, Fisher's exact test (primary care versus hospital or perinatal unit), unadjusted. Additional summary (analysis file, n = 1331): 97 respondents (7.3%) selected all three relevant orders and no distractor, they recognized a mean of 2.0 of five diagnostic methods; 79 (5.9%) named all five.

## Table S2. Checklist for Reporting Results of Internet E-Surveys (CHERRIES)

| Item category | Checklist item | Reported |
|---|---|---|
| Design | Describe survey design | Cross-sectional open online survey; convenience sample of clinical staff in antenatal care (Section 2.1–2.2). |
| IRB approval and informed consent | IRB approval | Local Bioethics Commission, Khoja Akhmet Yassawi University, protocol No. 4/26 (8 May 2026), conclusion No. 216 (29 May 2026) (Section 2.6; S2). |
| | Informed consent | Opening page stated the purpose, intended use of data and anonymity; electronic consent required to proceed (item C1). |
| | Data protection | No names, contact details, facility identifiers or IP-based identifiers collected; data held by the study team. |
| Development and pre-testing | Development and testing | Items mapped to national orders; reviewed for clarity by two obstetrician-gynecologists and one public health specialist (Section 2.3). No formal pilot. |
| Recruitment process | Open or closed survey | Open survey; link distributed through the regional health administration. |
| | Contact mode | Initial contact online through regional health administration channels. |
| | Advertising | No public advertising; invitation by regional health administration only. |
| Survey administration | Web/E-mail | Web form (Google Forms); data captured automatically. |
| | Context | Invitation distributed to facilities of all levels in Turkistan region. |
| | Mandatory/voluntary | Voluntary participation. |
| | Incentives | None. |
| | Time/date | 1 June to 30 July 2026. |
| | Randomisation of items | No; fixed order in four blocks. |
| | Adaptive questioning | No. |
| | Number of items | 38 items (35 content items, 3 respondent items). |
| | Number of screens | Four content blocks after the consent and respondent items. |
| | Completeness check | All items mandatory; a form could be submitted only when complete. |
| | Review step | Not specified in the form settings reported here. |
| Response rates | Unique site visitor | Not recorded by the platform; view rate cannot be calculated. |
| | View rate | Not available. |
| | Participation rate | Not available (views not recorded). |
| | Completion rate | 1331 forms submitted; all complete (mandatory items). |
| Preventing multiple entries | Cookies used | No. |
| | IP check | No (IP addresses not collected). |
| | Log file analysis | Timestamps checked: all 1331 distinct. |
| | Registration | No registration; 13 repeated full response profiles (19 additional records) retained in the primary analysis and removed in a sensitivity analysis. |
| Analysis | Handling of incomplete questionnaires | Not applicable (no incomplete submissions). |
| | Questionnaires submitted with an atypical timestamp | Completion time not recorded; not assessed. |
| | Statistical correction | No weighting; sample described as a convenience sample; no regional workforce denominator was obtained, so coverage cannot be estimated. |

Checklist from Eysenbach (J. Med. Internet Res. 2004, 6, e34).

## Table S3. Knowledge items by level of care

| Code | Item | Correct, n (%) N = 1331 | 95% CI | Primary care, % correct | Hospital or perinatal, % correct | p | Most frequent incorrect answer, n (%) |
|---|---|---|---|---|---|---|---|
| K1 | What are the main aims of prenatal genetic screening? | 1224 (92.0) | 90.4–93.3 | 92.1 | 91.1 | 0.768 | Determining fetal sex, 55 (4.1) |
| K2 | Which stages of prenatal screening during pregnancy are the most important? | 1195 (89.8) | 88.0–91.3 | 89.8 | 89.4 | 0.224 | Third trimester, 73 (5.5) |
| K3 | What is karyotyping? | 836 (62.8) | 60.2–65.4 | 61.7 | 69.4 | 0.147 | A method for detecting gene mutations, 243 (18.3) |
| K4 | Which first-trimester maternal serum biochemical markers are used in Kazakhstan to detect developmental defects and chromosomal anomalies? | 776 (58.3) | 55.6–60.9 | 59.6 | 50.6 | 0.075 | PAPP-A, β-hCG, AFP, 477 (35.9) |
| K5 | Trisomy (Down syndrome) is associated with an anomaly of which chromosome? | 772 (58.0) | 55.3–60.6 | 56.6 | 67.2 | 0.022 | X chromosome, 269 (20.2) |
| K6 | Trisomy (Patau syndrome) is associated with an anomaly of which chromosome? | 758 (56.9) | 54.3–59.6 | 56.3 | 61.1 | 0.763 | Chromosome 18, 205 (15.4) |
| K7 | Trisomy (Edwards syndrome) is associated with an anomaly of which chromosome? | 686 (51.5) | 48.9–54.2 | 51.2 | 53.9 | 0.784 | Chromosome 13, 247 (18.6) |
| K8 | In which week of pregnancy is the first prenatal ultrasound screening performed? | 652 (49.0) | 46.3–51.7 | 50.6 | 38.3 | <0.001 | $9^{+0}$–$12^{+6}$ weeks, 350 (26.3) |
| K10 | In which week of pregnancy is the second prenatal ultrasound screening performed? | 483 (36.3) | 33.7–38.9 | 36.9 | 32.8 | 0.007 | $18^{+0}$–$21^{+6}$ weeks, 401 (30.2) |
| K11 | Which examinations make up combined prenatal screening in the first trimester? | 483 (36.3) | 33.7–38.9 | n/a | n/a | n/a | Ultrasound alone, 292 (21.9) |
| K12 | In what unit are the results of first-trimester (combined) prenatal screening interpreted? | 218 (16.4) | 14.5–18.5 | 13.4 | 35.6 | <0.001 | IU/L, 499 (37.5) |
| K9 | From what age are pregnant women considered at high risk of congenital and hereditary pathology? | 148 (11.1) | 9.5–12.9 | 9.8 | 19.4 | 0.003 | From 35 years, 550 (41.4) |

Proportion correct from the analysis file (n = 1331); K9 is shown against the national key (37 years). p, Fisher's exact test across all response options, primary care versus hospital or perinatal unit (export, n = 1330). K11 was scored all-or-nothing from a multiple-response list, so workplace percentages are not available from the export.

## Table S4. Item responses by level of care

Primary care versus hospital or perinatal unit (institutional export; Fisher's exact test, unadjusted).

| Response | Primary care, n (%) N = 1150 | Hospital or perinatal, n (%) N = 180 |
|---|---|---|
| **AW. Do you know whether a medical genetics service operates in the region where you work? (p = 0.059)** | | |
| Yes, fully | 756 (65.7) | 134 (74.4) |
| Partly | 237 (20.6) | 30 (16.7) |
| Not aware | 157 (13.7) | 16 (8.9) |
| **K1. What are the main aims of prenatal genetic screening? (p = 0.768)** | | |
| Assessing the risk of chromosomal anomalies | 1059 (92.1) | 164 (91.1) |
| Determining fetal sex | 46 (4.0) | 9 (5.0) |
| Assessing placental function | 45 (3.9) | 7 (3.9) |
| **K8. In which week of pregnancy is the first prenatal ultrasound screening performed? (p = <0.001)** | | |
| $9^{+0}$–$12^{+6}$ weeks | 301 (26.2) | 49 (27.2) |
| $10^{+0}$–$14^{+6}$ weeks | 118 (10.3) | 19 (10.6) |
| $11^{+0}$–$13^{+6}$ weeks | 582 (50.6) | 69 (38.3) |
| $12^{+0}$–$14^{+6}$ weeks | 149 (13.0) | 43 (23.9) |
| **K10. In which week of pregnancy is the second prenatal ultrasound screening performed? (p = 0.007)** | | |
| $16^{+0}$–$20^{+0}$ weeks | 183 (15.9) | 32 (17.8) |
| $18^{+0}$–$21^{+6}$ weeks | 331 (28.8) | 70 (38.9) |
| $19^{+0}$–$21^{+0}$ weeks | 424 (36.9) | 59 (32.8) |
| $19^{+6}$–$21^{+0}$ weeks | 212 (18.4) | 19 (10.6) |
| **K4. Which first-trimester maternal serum biochemical markers are used in Kazakhstan to detect developmental defects and chromosomal anomalies? (p = 0.075)** | | |
| PAPP-A, β-hCG | 685 (59.6) | 91 (50.6) |
| PAPP-A, β-hCG, AFP | 401 (34.9) | 76 (42.2) |
| Inhibin A | 20 (1.7) | 6 (3.3) |
| Unconjugated estradiol | 44 (3.8) | 7 (3.9) |
| **K3. What is karyotyping? (p = 0.147)** | | |
| A method for studying chromosome number and structure | 710 (61.7) | 125 (69.4) |
| A method for detecting gene mutations | 213 (18.5) | 30 (16.7) |
| Assessing fetal developmental stage by ultrasound | 174 (15.1) | 17 (9.4) |
| Assessing hormone levels in the body | 53 (4.6) | 8 (4.4) |
| **K9. From what age are pregnant women considered at high risk of congenital and hereditary pathology? (p = 0.003)** | | |
| From 30 years | 161 (14.0) | 20 (11.1) |
| From 32 years | 34 (3.0) | 2 (1.1) |
| From 35 years | 486 (42.3) | 64 (35.6) |
| From 37 years | 113 (9.8) | 35 (19.4) |
| From 40 years | 356 (31.0) | 59 (32.8) |
| **K12. In what unit are the results of first-trimester (combined) prenatal screening interpreted? (p = <0.001)** | | |
| MoM (multiple of the median) | 154 (13.4) | 64 (35.6) |
| IU/L | 447 (38.9) | 52 (28.9) |
| g/L | 260 (22.6) | 30 (16.7) |
| "MM" (abbreviation as printed on the form) | 289 (25.1) | 34 (18.9) |
| **A2. Do you think prenatal screening for fetal malformations and chromosomal anomalies should be mandatory for all pregnant women? (p = 0.103)** | | |
| Yes, mandatory | 970 (84.3) | 156 (86.7) |
| Yes, recommended | 101 (8.8) | 8 (4.4) |
| Only when indicated | 53 (4.6) | 13 (7.2) |
| No | 26 (2.3) | 3 (1.7) |

| Response | Primary care, n (%) N = 1150 | Hospital or perinatal, n (%) N = 180 |
|---|---|---|
| **A1. What is your attitude to referring pregnant women at risk of congenital and hereditary pathology to a geneticist? (p = 0.127)** | | |
| I consider it mandatory | 1004 (87.3) | 157 (87.2) |
| Recommended but not mandatory | 97 (8.4) | 9 (5.0) |
| Not needed, should be voluntary | 27 (2.3) | 11 (6.1) |
| Negative | 22 (1.9) | 3 (1.7) |
| **TOP. In your opinion, is termination of pregnancy on medical grounds appropriate when a severe untreatable hereditary disease is detected? (p = 0.141)** | | |
| Yes | 898 (78.1) | 131 (72.8) |
| Difficult to answer | 209 (18.2) | 44 (24.4) |
| No | 43 (3.7) | 5 (2.8) |
| **PAW. How do you rate the population's awareness of what medical genetics services offer? (p = 0.532)** | | |
| High | 513 (44.6) | 84 (46.7) |
| Moderate | 554 (48.2) | 80 (44.4) |
| Low | 83 (7.2) | 16 (8.9) |
| **AC. How do you rate the accessibility of medical genetic care for pregnant women in your region? (scale 1–5) (p = 0.013)** | | |
| 1 – Not accessible | 132 (11.5) | 17 (9.4) |
| 2 – Accessible with difficulty | 103 (9.0) | 7 (3.9) |
| 3 – Partly accessible | 180 (15.7) | 27 (15.0) |
| 4 – Generally accessible | 434 (37.7) | 62 (34.4) |
| 5 – Fully accessible | 301 (26.2) | 67 (37.2) |
| **A5. In your opinion, is invasive prenatal diagnosis necessary when indicated? (p = 0.901)** | | |
| Necessary, mandatory | 1061 (92.3) | 169 (93.9) |
| With the pregnant woman's consent (free text) | 3 (0.3) | 0 (0.0) |
| Not necessary | 80 (7.0) | 11 (6.1) |
| I do not know (free text) | 6 (0.5) | 0 (0.0) |
| **A4. Do you think developing medical genetics services is important for the country? (p = 0.348)** | | |
| Yes, very important | 835 (72.6) | 138 (76.7) |
| Yes, important | 265 (23.0) | 33 (18.3) |
| Difficult to answer | 40 (3.5) | 6 (3.3) |
| No, not important | 10 (0.9) | 3 (1.7) |
| **A6. How confident are you in your knowledge of prenatal genetic screening for fetal malformations and chromosomal anomalies? (p = 0.483)** | | |
| Fully confident | 441 (38.3) | 76 (42.2) |
| I know the main aspects | 218 (19.0) | 33 (18.3) |
| I need further training | 448 (39.0) | 68 (37.8) |
| I do not understand the topic | 43 (3.7) | 3 (1.7) |
| **A3. Do you think introducing genetic counselling reduces the birth rate of children with hereditary diseases? (p = 0.641)** | | |
| Yes | 1012 (88.0) | 157 (87.2) |
| Maybe (free text) | 8 (0.7) | 0 (0.0) |
| No | 129 (11.2) | 23 (12.8) |
| Did not understand (free text) | 1 (0.1) | 0 (0.0) |
| **P7. In the past year, have you referred patients to a medical geneticist for consultation? (p = <0.001)** | | |
| Frequently | 507 (44.1) | 62 (34.4) |
| Sometimes | 381 (33.1) | 53 (29.4) |
| Did not refer | 262 (22.8) | 65 (36.1) |
| **P5. Do you use the results of medical genetic tests in your practice? (p = 0.004)** | | |
| Yes, regularly | 571 (49.7) | 70 (38.9) |
| Sometimes | 409 (35.6) | 68 (37.8) |
| Never | 170 (14.8) | 42 (23.3) |
| **P4. Do you talk with patients about the importance of prenatal and neonatal screening? (p = 0.060)** | | |

| Response | Primary care, n (%) N = 1150 | Hospital or perinatal, n (%) N = 180 |
|---|---|---|
| Frequently | 643 (55.9) | 93 (51.7) |
| Sometimes | 401 (34.9) | 59 (32.8) |
| Never | 103 (9.0) | 28 (15.6) |
| Of course (free text) | 3 (0.3) | 0 (0.0) |
| **P1. Do you recommend molecular genetic testing to patients when indicated? (p = 0.428)** | | |
| Yes | 847 (73.7) | 138 (76.7) |
| Sometimes | 218 (19.0) | 27 (15.0) |
| No | 85 (7.4) | 15 (8.3) |
| **SRC. Where do you get information about prenatal screening during pregnancy?** | | |
| Clinical guidelines | 589 (51.2) | 78 (43.3) |
| Scientific conferences or seminars | 152 (13.2) | 36 (20.0) |
| Colleagues | 226 (19.7) | 27 (15.0) |
| Internet or social media | 179 (15.6) | 33 (18.3) |
| "Always" (free text) | 3 (0.3) | 3 (1.7) |
| "Sometimes" (free text) | 1 (0.1) | 1 (0.6) |
| "Polyclinic" (free text) | 0 (0.0) | 1 (0.6) |
| "From the district nurse" (free text) | 0 (0.0) | 1 (0.6) |
| **P2. Do you talk with couples planning a pregnancy about the risk of hereditary diseases? (p = <0.001)** | | |
| Yes, regularly | 826 (71.8) | 107 (59.4) |
| Sometimes | 262 (22.8) | 47 (26.1) |
| No | 62 (5.4) | 26 (14.4) |
| **P6. How often do you receive patients' molecular genetic test results (e.g., when a hereditary disease is suspected)? (p = 0.028)** | | |
| Frequently | 510 (44.3) | 84 (46.7) |
| Rarely | 494 (43.0) | 62 (34.4) |
| Never | 146 (12.7) | 34 (18.9) |
| **P3. Do you advise patients to undergo NIPT (non-invasive prenatal testing) when indicated? (p = 0.330)** | | |
| Yes, always | 735 (63.9) | 109 (60.6) |
| Sometimes | 334 (29.0) | 53 (29.4) |
| Never | 81 (7.0) | 18 (10.0) |
| **TR. Have you attended training or seminars on medical genetics in the last 3 years? (p = 0.013)** | | |
| Yes | 364 (31.7) | 74 (41.1) |
| No | 786 (68.3) | 106 (58.9) |
| **K5. Trisomy (Down syndrome) is associated with an anomaly of which chromosome? (p = 0.022)** | | |
| Chromosome 21 | 651 (56.6) | 121 (67.2) |
| Chromosome 13 | 189 (16.4) | 16 (8.9) |
| Chromosome 18 | 72 (6.3) | 9 (5.0) |
| X chromosome | 235 (20.4) | 34 (18.9) |
| No answer | 3 (0.3) | 0 (0.0) |
| **K7. Trisomy (Edwards syndrome) is associated with an anomaly of which chromosome? (p = 0.784)** | | |
| Chromosome 18 | 589 (51.2) | 97 (53.9) |
| Chromosome 13 | 212 (18.4) | 35 (19.4) |
| Chromosome 21 | 173 (15.0) | 26 (14.4) |
| X chromosome | 172 (15.0) | 22 (12.2) |
| No answer | 4 (0.3) | 0 (0.0) |
| **K6. Trisomy (Patau syndrome) is associated with an anomaly of which chromosome? (p = 0.763)** | | |
| Chromosome 13 | 648 (56.3) | 110 (61.1) |
| Chromosome 18 | 180 (15.7) | 25 (13.9) |
| Chromosome 21 | 166 (14.4) | 24 (13.3) |
| X chromosome | 152 (13.2) | 21 (11.7) |

| Response | Primary care, n (%) N = 1150 | Hospital or perinatal, n (%) N = 180 |
|---|---|---|
| No answer | 4 (0.3) | 0 (0.0) |
| **K2. Which stages of prenatal screening during pregnancy are the most important? (p = 0.224)** | | |
| First and second trimester | 1033 (89.8) | 161 (89.4) |
| Third trimester | 66 (5.7) | 7 (3.9) |
| Only before delivery | 20 (1.7) | 7 (3.9) |
| Screening is not needed during pregnancy | 31 (2.7) | 5 (2.8) |

Multiple-response items are in Table S1. Comparisons are unadjusted and exploratory; with 30 tests, about 1.5 would be expected to reach $p < 0.05$ by chance.

## Table S5. Former attitude composite

| Measure | Attitude composite, 6 items (0–12) |
|---|---|
| Items | A1–A6 coded 2/1/0 (Supplementary Material S1) |
| Mean ± SD (% of maximum) | 9.96 ± 1.96 (83.0) |
| Median (interquartile range) | 10 (9–11) |
| Favourable (≥80% of maximum), n (%) | 906 (68.1) |
| Cronbach's alpha | 0.647 |
| KMO; eigenvalues > 1 | 0.742; 1 |
| aOR for recent training, favourable attitude (95% CI) | 1.67 (1.26–2.21) |

n = 1331. The composite is not used as an outcome, because its coding ranked "mandatory" above "recommended", it mixes confidence (A6) with attitudes, and it has a ceiling effect. It is retained only as a covariate in the practice models of the main text.

## Table S6. Selected exploratory cross-tabulations

Column percentages; p as reported in the institutional cross-tabulation (n = 1330). These comparisons were not pre-specified and are not adjusted for multiple testing or confounding.

**a. Self-rated confidence (A6) by genetics training in the previous 3 years (TR)** (p <.001)

| Confidence \ Training | No | Yes | Total |
|---|---|---|---|
| Knows the main aspects | 175 (19.6%) | 76 (17.4%) | 251 (18.9%) |
| Does not understand the topic | 38 (4.3%) | 8 (1.8%) | 46 (3.5%) |
| Fully confident | 246 (27.6%) | 271 (61.9%) | 517 (38.9%) |
| Needs further training | 433 (48.5%) | 83 (18.9%) | 516 (38.8%) |
| Total | 892 (100.0%) | 438 (100.0%) | 1330 (100.0%) |

**b. Importance of developing genetics services (A4) by training (TR)** (p = 0.001)

| Importance \ Training | No | Yes | Total |
|---|---|---|---|
| Difficult to answer | 33 (3.7%) | 13 (3.0%) | 46 (3.5%) |
| No, not important | 11 (1.2%) | 2 (0.5%) | 13 (1.0%) |
| Yes, important | 225 (25.2%) | 73 (16.7%) | 298 (22.4%) |
| Yes, very important | 623 (69.8%) | 350 (79.9%) | 973 (73.2%) |
| Total | 892 (100.0%) | 438 (100.0%) | 1330 (100.0%) |

**c. Awareness of a local genetics service (AW) by referral to a geneticist in the past year (P7)** (p <.001)

| Awareness \ Referral | Frequently | Did not refer | Sometimes | Total |
|---|---|---|---|---|
| Partly aware | 68 (12.0%) | 100 (30.6%) | 99 (22.8%) | 267 (20.1%) |
| Fully aware | 447 (78.6%) | 163 (49.8%) | 280 (64.5%) | 890 (66.9%) |
| Not aware | 54 (9.5%) | 64 (19.6%) | 55 (12.7%) | 173 (13.0%) |
| Total | 569 (100.0%) | 327 (100.0%) | 434 (100.0%) | 1330 (100.0%) |

**d. Awareness of a local genetics service (AW) by perceived accessibility, 1–5 (AC)** ($p < .001$)

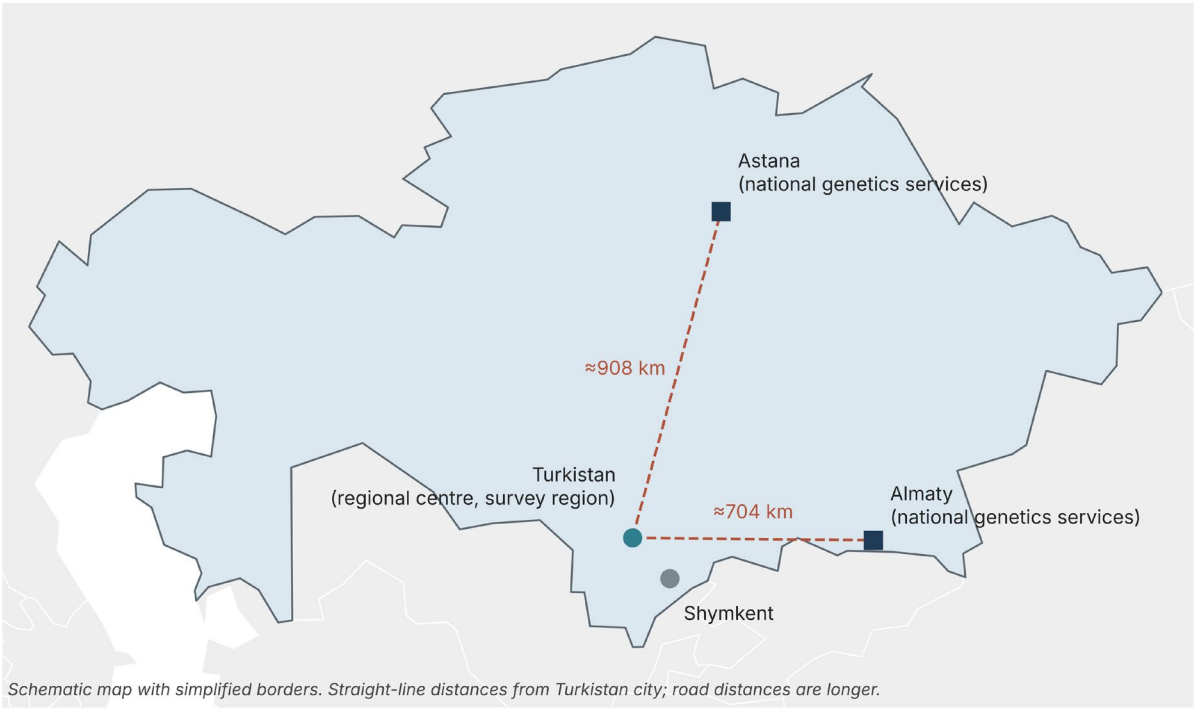
| Awareness \ Accessibility | 1 | 2 | 3 | 4 | 5 | Total |
|---|---|---|---|---|---|---|
| Partly aware | 23 (15.4%) | 32 (29.1%) | 78 (37.7%) | 98 (19.8%) | 36 (9.8%) | 267 (20.1%) |
| Fully aware | 93 (62.4%) | 49 (44.5%) | 93 (44.9%) | 343 (69.2%) | 312 (84.8%) | 890 (66.9%) |
| Not aware | 33 (22.1%) | 29 (26.4%) | 36 (17.4%) | 55 (11.1%) | 20 (5.4%) | 173 (13.0%) |
| Total | 149 (100.0%) | 110 (100.0%) | 207 (100.0%) | 496 (100.0%) | 368 (100.0%) | 1330 (100.0%) |

**e. Unit chosen for interpreting combined-screening results (K12) by use of genetic test results (P5)** ($p = 0.036$)

| Unit \ Use of results | Never | Yes, regularly | Sometimes | Total |
|---|---|---|---|---|
| IU/L | 69 (32.5%) | 253 (39.5%) | 177 (37.1%) | 499 (37.5%) |
| "MM" | 54 (25.5%) | 170 (26.5%) | 99 (20.8%) | 323 (24.3%) |
| MoM | 40 (18.9%) | 99 (15.4%) | 79 (16.6%) | 218 (16.4%) |
| g/L | 49 (23.1%) | 119 (18.6%) | 122 (25.6%) | 290 (21.8%) |
| Total | 212 (100.0%) | 641 (100.0%) | 477 (100.0%) | 1330 (100.0%) |

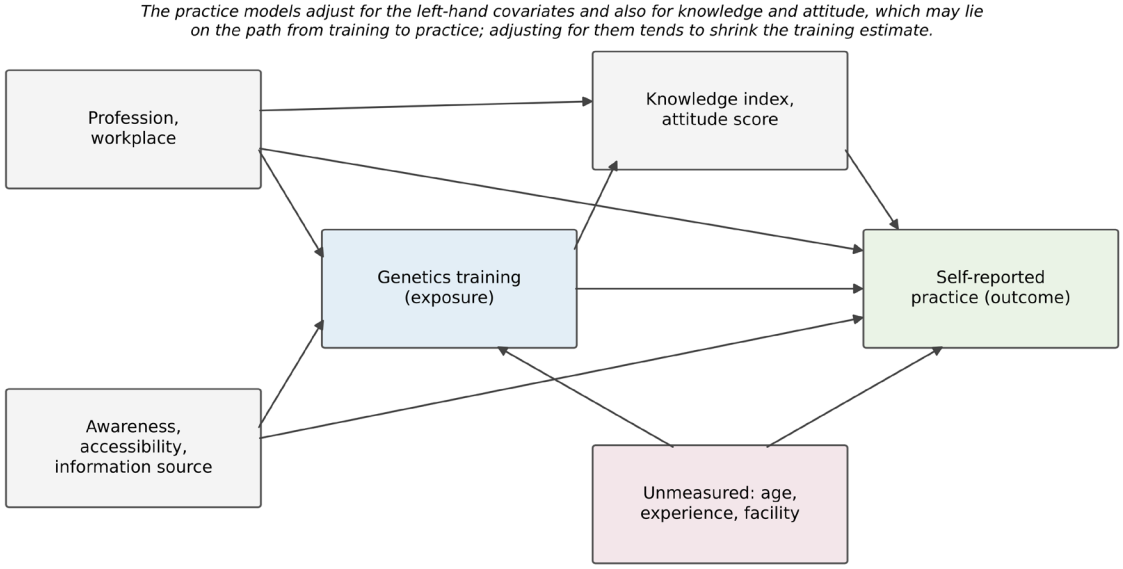
Among respondents trained in the previous three years, 61.9% described themselves as fully confident in their knowledge, compared with 27.6% of those not trained (Table S6a), although trained and untrained respondents did not differ in objective knowledge (main text, Table 6).

## Figure S1. Location of Turkistan region relative to the national medical genetics services




Schematic map with simplified borders. Dashed lines show straight-line distances from Turkistan city to Almaty (≈704 km) and Astana (≈908 km); road distances are longer.

## Figure S2. Directed acyclic graph for the practice models




Training is the exposure and self-reported good practice the outcome. The practice models adjust for profession, workplace, awareness, accessibility and information source, and also for the knowledge index and the former attitude score. These two may lie on the path from training to practice, so adjusting for them tends to shrink the training estimate. Age, experience and facility were not recorded and remain unmeasured confounders.