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Tilmaanta Baaritaanka Uur Jiifka

In aad wax ka baratid caafimaadka cunugaada

Cutubkaan wuxuu sharaxaa baarista uur jiifka ee macluumaad ka bixiya caafimaadka cunugaada. Adiga ayaa doorashada iska leh oo ay kugu xidhantahay in aad doonaysid in laguu sameeyo baadhitaanadan iyo in kale. La hadal daryeelahaaga caafimaad si wax dheeraad ah u kaa ogaado/barto iyo in u kaa caawiyo go'aan qaadashada in ay wax baadhitaanadani ka mid ihi ay kugu haboon yihiin.

Haddii laguu sameeyo mid ka mida ah baaritaankaan, waxaa lagu weydiin doonaa in aad in badan ka akhrisid mid kasta. Waxaa lagu weydiin doonaa in aad akhrisid iyo in aad baaritaan kasta u saxiixdid foomka ogolaashada.

Waxyaabo badan baad samayn kartaa marka aad uurka leedahay si aad u ilaaliso caafimaadkaaga iyo ka ilmahaaga. Qaadashada fiitamiinada dhalmada ka hor, cunidda cuntooyin caafimaad qaba, jimicsi, iyo hurdo kugu filan dhamaan waa muhiim.

Jidhka aadanuhu waa adagyahay. Xitaa haddii aad u samayso wax kasta "sax" inta aad uurka leedahay, ubadku badiyaaba si caadi ah uma samaysmaan/horumaraan. Boqolkiiba inta u dhexeyso 3% iyo 5% caruurta (inta u dhexeyso 3 iyo 5 marka la soo qaado 100) waxay qabaan nooc ka mid ah cilladaha caafimaadka marka ay dhashaan.

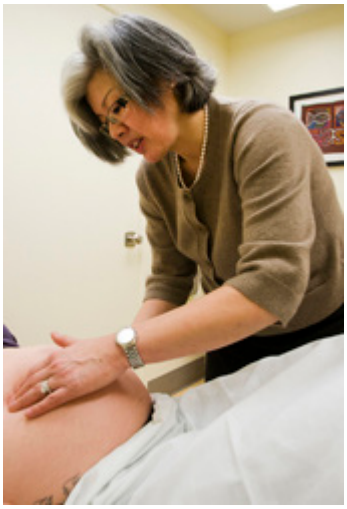
Waraaqahan/handout waxay ku siinayaan macluumaad asaasi ah oo ku saabsan badhitaanada si ay kaaga gacansiiyaan ka go'aan gaadhista ta kuu wanaagsan.

Maxay yihiin baadhitaanadani?

Waxa jira laba nooc oo baadhitaano oo aasaasi ah:

- **Baadhitaanada screeningka** waxay saadaaliyaan oo ay sii sheegaan *nasiibka*, ama masiibooyinka, in u ilmahaagu leeyahay cilad dhalasho oo gaar ah.
- **Baadhitaanada lafagurka** waxay sheegaan haddii ilmahaagu laayahay ama aanu lahayn cilad dhalasho oo gaar ah.

Jadwalada ku yaal bogga xiga waxaa ku qoran liiska baaritaanada iyo marka la sameeyo. Waxay kaloo ku siiyaan sharaxaad kooban oo ku saabsan baaritaan kasta iyo waxa ay kuu sheegi doonto. Inta ka hartay cutubkaan wuxuu ku siiyaa tafaasiil badan oo ku saabsan baaritaanka, haddii aad rabtid in aad wax ka akhrisatid ka hor inta aadan la hadlin bixiyahaada xanaanada caafimaadka.



Baarista ka horeeyso dhalashada waxay macluumaad ka bixin karaan caafimaadka cunugaada.

Baadhitaanada Screeningka ah

Magaca Baadhitaanka ah	Goorma	Sharaxa	Maxay Kuu Sheegaysaa
Nuchal translucency (NT) ultrasound	11 ilaa 14 todobaad	Sawiraada Bogga/ Abdominal ultrasound si loo qiyaaso/ cabiro inyar oo banaan oo ilmaha deligiisa ah.	*Inta* ay llegtahay dhibaatada ilmaha hedesidihiisu (chromosome problem).
Waxyaabaha wax lagu eego/arko oo iskudhafa (Integrated screen)	11 ilaa 14 todobaad *iyo* 15 ilaa 22 todobaad	NT ultrasound ku dar 2 nooc (sample) oo dhiig ah oo kala soocan.	*Khataraha* u ilmahaagu u lahaan karo Down syndrome, trisomy 18, ama ciladda laf dhabarka (spina bifida).
Quad screen	15 ilaa 22 todobaad	1 nooc oo dhiig ah.	*Khataraha* u ilmahaagu u lahaan karo Down syndrome, trisomy 18, ama ciladda laf dhabarka (spina bifida).

Baadhitaano Lafagur

Magaca Baadhitaanka ah	Goorma	Sharaxa	Maxay Kuu Sheegaysaa
Qaadidda Kambiyoonaha Ibada (Chorionic villus sampling) (CVS)	11 ilaa 14 todobaad	Muunad madheerta ah (Sample of placenta) oolaga soo qaadayo xubnaha taranka am uur ku jirta.	*Inuu ilmahaagu leeyahay dhibaato* iyo in kale iyo mar mar cudurada la iska dhaxlo.
Dhacaanka laga qaado uur jiifka [Amniocentesis (lagu qaado ultrasound)]	16 ilaa 22 todobaad	Nooc dhiiga oo jidhka ilmahaaga laga soo wada qaado.	*Inuu ilmahaagu leeyahay dhibaato* chromosome iyo in kale, ciladda laf dhabarka (spina bifida), iyo mararka qaarkood cuduro la iska dhaxlo ee kale.

Badhitaano Kale

Magaca Baadhitaanka ah	Goorma	Sharaxa	Maxay Kuu Sheegaysaa
Koriimada Uur Jiifka lagu ogaado ultrasound (Anatomy ultrasound)	18 ilaa 22 todobaad	Baadhitaanka bogga dhexdiisa si loo hubiyo koritaanka iyo horumarka ilmaha.	*In xaalado aan caadi* ahayn looga shikiyey iyo in kale iyo haddii baadhis loo baahan yahay.

Baadhitaanada Screeningka

Nuchal Translucency ama NT Ultrasound

Badhitaankan screeningkat waxa lagu sameeyaa uurka 11 ilaa 14 todobaad dhexdooda. Iyadoo la adeegsanayo qalabka bogga lagu baadho, waxa la qiyaasayaa/cabirayaa dherarka ilmaha si u waafaqo soo gelitaanka taariikhdaada. Sawiraada Uurkujirka dhexdiisa/Abdominal ultrasound waxa walibana loo adegsadaa si loo qiyaaso/cabiro inyar oo banaan oo ah ilmaha deligiisa ah. Intan banana waxa loo yaqaan *nuchal translucency* (NT). Weynaanta inta u dareeruhu ku jiro ayey ku xidhantahay *khatarta/chanceska* u ilmahaagu u u leeyahayay dhibaatada chromosomeka. Ultrasoundka NT waxaa samayn kara oo keliya shaqaale si gaar ah loo tababaray.

Integrated screen

Baaritaankaan wuxuu isticmaalaa natiijadaha ka soo baxa ultrasoundka NT iyo 2 baaritaanada dhiigga. Kambiyoonaha dhiigga kowaad waxaa la qaadaa inta u dhexeyso 11 iyo 14 todobaad, badanaa isla maalinta la qaado ultrasoundka NT. Kambiyoonaha dhiigga 2aad waxaa la qaadaa inta u dhexeyso 15 iyo 22 todobaad. Baadhitaanadu waxay raadiyaan qaabab/noocyo borotiin ah iyo isirsidayaal (hormones) kuwaasoo ku xidhan ama xidhiidh la leh cilado dhalasho oo gaar ah.

Screenada isudhafan waxay ku sheegayaan *mararka* u ilamahaagu yeelanaayo ama ay heli karaan Down syndrome, trisomy 18, ama ciladda laf dhabarka (spina bifida). (Fiiri tiirarka dhinacyada ku yaal bogagga 34, 35, iyo 36 si aad u heshid tafaasiil badan.) Ma baarto xaaladahaan. Haweenka la mariyo screenada isudhafan ee aan caadiga ahayn natiijadiisu badankoodu waxay muujinayaan in ay wali haystaan ilmo caafimaad qaba.

Muraayadda midaysan (integrated screen) waxay arki kartaa:

- 90 marka la soo qaado 100 kiis (90%) calaamadda Barnimada (Down syndrome)
- 90 marka la soo qaado 100 kiis (90%) trisomy 18 (itaal darrida xubnaha qaar)
- 80 marka la soo qaado 100 kiis (80%) ciladda laf dhabarka (spina bifida) (cilladda laf dhabarka)

Hase ahatee, **ma** arki doonto dhamaan kiisaska cilladahaan dhalashada. Iyo ma baarto cillado kale oo la xariira caafimaadka.

Quad Screen

Baaristaan waxay ku lug leedahay 1 kambiyoonaha dhiiga oo la qaado inta u dhexeyso 15 ilaa 22 todobaad. Wuxuu la mid yahay screenka isudhafan, waayo waliba waxay raadisaa qaabab/noocyo borotiin iyo hormones ah kuwaasoo xidhiidh la leh ciladooyin dhalasho oo gaar ah.

Screenka banaanku wuxuu ku sheegayaa *mararka* ilmahaagu leeyahay Down syndrome, trisomy 18, ama ciladda laf dhabarka (spina bifida). Ma lafagurayo xaaladahan. Haweenka hela xaaladaha aan caadiga ahayn ee natiijada screenka banaanka waxay wali haystaan ilmo caafimaad qaba.

Waa maxay dhibaatada chromosome?

Chromosomesku waa qaybo ka kooban tilmaamaha iyo macluumaad hideside. Waxaan ka dhaxalnaa waalidiintena. Waxay kantaroolaan koriimada jirkena iyo horumarkiisa. Qayb kasta oo jidhkeena ah bay ka mid yihiin, waxaana lagu arkaa qalabka wax weyneeya (microscope) oo kaliya.

Dadka badankiisu waxay leeyihiin 46 chromosomes, hase ahaatee qaar baa leh chromosomeyo dheeraad ah (47), mid maqan (45), ama chromosome badh/jeex ka jaban yahay ama mid badh/jeex dheeraad ahii ku dhegan yahay.

Isbedel ku dhacaa chromosomeka badiyaaba wuxuu sababaa cilad dhalasho iyo habsan/dib-u-dhac horumar, hase ahaatee ma aha arrin marwalba dhacda.

Baarista "quad" waxaa lagu arkaa:

- 85 marka la soo qaado 100 kiis (85%), calaamadda Barnimada (Down syndrome)
- 75 marka la soo qaado 100 kiis (75%), trisomy 18 (itaal darrida xubnaha qaar)
- 80 marka la soo qaado 100 kiis (80%) , cilladda laf dhabarka (spina bifida)

Hase ahatee, **ma** ogaan doonto dhamaan kiisaska cilladaha dhalmada. Iyo, ma baarto dhibaato kasta oo la xariirto caafimaadka.

Waxaa dhici karto in baarista "quad" fiican tahay haddii aadan bilaabin daryeelka uur jiifka ilaa aad ka gaartid bisha 4aad ama haddii aan la heli karin ultrasoundka NT.

Baarista "Advanced Aneuploidy" ee Lagu Sameeyo DNAda ka Madax Banaan Unugga

Waxaa dhici karto in aad wararka ka maqashay ama aad internetka ku aragtay qoraalo ku saabsan baaritaan cusub oo lagu ogaan karo calaamadda Barnimada (Down syndrome). Baaristaan waxaa lagu magacaabaa *"advanced aneuploidy screening with cell-free DNA"*. Waxay isticmaashaa kambiyoone laga qaado dhiigga hooyada, waxaana la sameyaa marka la gaaro 10 todobaad oo xaamilonimo ah. Waxay baartaa qaladaadka qaar ee ku dhaca kromosoomka cunugga.

Qof kasta wuxuu qabaa xoogaa DNA madax banaan (aan ku jirin gudaha unugga) oo ku jirta dhiigga. Marka aad xaamilo noqotid, inta badan DNAda madax banaan waxay kaa yimadaan adiga, hase ahatee qaar waxay ka yimadaan xaamilonimadaada. Baaristaan, waxaa la cabbiraa wadarta qiyaasta DNAda ka madax banaan unugga ee ka yimaada kromosoomka 21, 18 iyo 13, kuna dhex jira dhiigaada.

Sida baarista kale, baaristaan kuuma sheegto haddii cunugga uu qabo, ama haddii uusan qabin, dhibaato xagga kromosoomka. Hase ahatee, haddii uu kor u kaco qiyaasta DNAda mid ka mid ah kromosoomyadaan ku jira dhiigaada, waxay u badan tahay in cunugga uu qabo "trisomy" la xariira kromosoomkaas.

Iminka, baaristaan waxaa lagu sameyaa dumarka qaba halista sare in ay qaadaan cunug qaba calaamadda Barnimada (Down), trisomy 18, trisomy 13. Haddii aad horay u dhashay cunug qaba mid ka mid ah trisomyadaan, ama haddii aad qaadatid baaritaan kale oo muujiya in natiijadaha aysan caadi ahayn, waxaa dhici karto in lagu siiyo baarista "aneuploidy screening" ee la xariirta DNAda ka madax banaan unugga.

Baadhitaanada Lafagurka ah

Anatomy Ultrasound

Baadhitaankan waxa la sameeyaa 18 iyo 22 todobaad dhexdood. Baadhis bogga waxa loo adeegsadaa in aad ku eegto ilmahaaga, inata dareera ah ee hareerihiisa, madheertaada, iyo ilmo galeenkaaga. Waxa lagu hubiyaa in u ilmuhu korayo iyo in halbowlayaasha waaweyn oo dhan ay samaysmeen.

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Muxuu yahay Down syndrome?

Down syndrome (calaamadda Barnimada) waxaa kaloo loo yaqaan trisomy 21. Waxaa lala kulmaa marka qof qabo koobi dheeraad ah kromosoomka 21.

Down syndrome wuxuu dadka u saameeyaa siyaabo kala duwan. Dadka qaba Down syndrome badiyaaba way ka muuqaal duwan yihiin xubnaha kale ee qoyskooda. Had iyo jeer waxay leeyihiin dib u dhac horumar/koriin, hase ahaatee heerka dib u dhucu qofba qofka kale wuu ka duwan yahay.

Dadka waaweyn/waayeelka ah ee qaba Down syndrome waxa laga yaabaa in ay ciyaari karaan isboorti/ciyaaro, shaqo aasaasi ah haysta, saaxiibana la maaweelan kara. Hase ahatee, badanaa ma noolaan karaan keligood haddii aysan helin kaalmo.

Caruur badan oo qabta Down syndrome waxay leeyihiin cilad wadnaha ah, kuwaasoo marmarka qaarkood lagu hagaajiyo qaliin. Dhibaatooyin kale oo caafimaad iyo cillado dhalasho marmar waxay la socdaan Down syndrome, hase ahaatee way yaryihiin.

Ilmahaagu in ku filan buu korey da'dan oo baadhista bogga (ultrasound) waxa laga yaabaa in u ka helo dhibaatooyin sida cilad beer xanuun oo ba'an, ciladda laf dhabarka (spina bifida), keli maqan, iyo dabin si ba'an u faruuran. Inkastoo baaritaankaan uusan sheegi doonin cilladaha kromosoomka, waxay muujin kartaa calaamadahooda ama xaalado kale.

Chorionic Villus Sampling ama CVS

Baaristaan lagu ogaanaayo cilladda waxaa badanaa la sameyaa inta u dhexeyso todobaadyada 11 iyo 14. Dhakhtarku wuxuu adeegsadaa irbad dhuuban oo dabacsan ama tuyuub/tuumbo dhuuban si u uga soo saaro muunad yar madheerta. Baadhitaan bogga ayaa la sameeyaa isla markaana, sidaas darteed ilmahaaga waa la arki karaa inta u habraacu socdo.

Muunada madheerta waxa loo adeegsadaa lafagurka dhibaatooyinka chromosomeka. Haddii ay xaalad la dhaxlo sida *"muscular dystrophy"* ama *"hemophilia"* ka jirto qoyskaada, kambiyoonaha waxaa loo adeegsan karaa in cunugaada looga baaro xaaladaas.

Fursadda in la dhiciyo kaddib marka la qaado CVS waa 1 marka la soo qaado 2 dumar, 100 (1% ilaa 2%).

Amniocentesis ama Amnio

Baaristaan lagu ogaanaayo cilladda jirta waxaa badanaa la qaadaa inta u dhexeyso 16 ilaa 22 todobaad. Dhaktarku wuxuu isticmaalaa irbad dhuubah oo jilicsan si u ugu qaado laba maalqaacadood oo dareera ku wareegsan ilmaha. Ultrasound ayaa isla markaaba lagu sameeya, sidaa awgeed ilmahaaga waa la arki karaa inta u habraacu socdoso.

Dareeraha waxa loo adeegsadaa in lagu lafaguro dhibaatooyinka chromosomeka iyo ciladda laf dhabarka (spina bifida). Haddi ay jirto xaalad la iska dhexlo sida (muscular dystrophy ama hemophilia) laga helo qoyskaagay, dareeraha waxa loo adeegsan karaa in lagu baadho ilmahaaga xaaladiisa.

Fursadaha lagu sameeyo amniocentesiska ee sababi doonaa dhicin waa 400ba 1 haweenay ah (0.25%).

Screeningka ku Salaysan Side la Kala Dhexlo

Tafiirtaada, ama isirkaaga, waa mid fure u ah ka gacansiinta barashada haddii ilmahaagu lahaan karo cudur la iska dhaxlo oo dhif ah. Koox kasta abtirsiinada waxay qabtaa xaalada la dhaxli karo oo ku badan kooxdaas marka la barbar dhigo kooxaha isirada kale. Xaaladaha ku xeran koox kasta oo ka tirsan isirka waxaa lagu qoray bogga 36.

Inta badan waqtiyada, labada qof waxay yeelan karaan cunug qaba mid ka mid ah cilladahaan marka *labada waalid* yahiin "siddeyaal" ("carriers") *isla* cilladaas. **Badanaa siddeyaasha ma laha calaamadaha cudurka.** Sidoo kale, inta badan siddeyaasha ma qabaan taariikh la xariirta qoyska oo muujisa cudurka. Haddii mid ka mid ah qoyskiina u leeyahay xaalad xaaladahan ka mid ah, u sheeg daryeel ku siiyahaaga caafimaad.

Waa maxay trisomy 18?

Trisomy 18 waxaa kaloo loo yaqaan calaamadda Edwards. Waxaa lala kulmaa marka qofka leeyahay koobi dheeraad kromosoomka nambarka 18.

Caruurta qabta trisomy 18 badankood ma noolaadaan uruurka. Caruurta qabta trisomy 18 waxay qabaan burbur ku yimaada maskaxda, iyo badanaa cillado kale, sida cilladaha wadnaha iyo cagta duuban.

Waa maxay cilladda laf dhabarka (spina bifida)?

Ciladda laf dhabarka (spina bifida) waa xaalad ay qayb ka mid ah xangulada dhabarka ilmaha aanay si caadi ah u samaysmin neerfayaasha xangulada ku jiraa ay cawarmaan/waxyeeloobaan. Arrintani waxay dhacdaa todobaadyada ugu horeeya uurka.

Ciladda laf dhabarka (spina bifida) waxay ku waxyeelaysaa dadka siyaabo kala duwan. Dad socodka ayey ku dhibtaa waxay u baahan karaan in ay adegsadaan cuskasho ama kursiga/baaskiilka naafada ee lagu socdo. Qaar waxa dhibi ka haysataa xakamaynta/ kantaroolka kaadihaysta. Marmarka qarkood, ciladda laf dhabarka (spina bifida) waxay sababi kartaa waxyeelo maskaxeed iyo dib u dhac koritaan/horumar.

Haddii adiga iyo saaxiibkaa aad labadaba tahiin siddeyaal isla xaaladda geneetikada, markaas waxaa dhici karta in cunugaada uu dhaxlo xaaladaas. Haddii aad xaqiiqa ahaan in aad u ogaato dhalashada ka hor, waxa la samayn karaa amniocentesis ama CVS. Muraayadda midaysan, muraayadda "quad", iyo ultrasoundka **ma** sheegi doonaan cilladahaan.

Si loo arko haddii aad tahay siddaha xaaladahaan dhaxalka, waxaad u baahan tahay in aad bixisid kambiyoone yar oo dhiig ah. Adiga ayay ku jirtaa haddii aad qaadaneysid qaar ka mid ah ama dhamaan baaristaan.

Jadwalkaan waxaa laga soo qaatay buugga "Ancestry-Based Carrier Screening" ("Baarista Siddaha Ku Saleysan Dhaxalka") waxaa daabacay Bulshada Qaranka La-Talliyeyaasha Geneetikada (National Society of Genetic Counselors, Inc.), 2005:

Meesha u ka soo Jeedo	**Xaaladda Dhaxalka**	**Inta u Side Ahaan Karo**
Maraykanka-Madow	Beta Thalassemia	10% (10 marka la soo qaado 100)
	Sickle Cell Disease	11% (11 marka la soo qaado 100)
Dadka Yurubta-Bari (Ashkenazi) Yuhuud "Jewish"	Canavan Disease	2.5% (2 ilaa 3 marka la soo qaado 100)
	Cystic Fibrosis	3% ilaa 4% (3 ilaa 4 marka la soo qaado 100)
	Familial Dysautonomia	3% (3 marka la soo qaado 100)
	Tay-Sachs Disease	3% (3 marka la soo qaado 100)
Rer-Yurub Caddaan	Cystic Fibrosis	3% (3 marka la soo qaado 100)
Mediterranean	Beta Thalassemia	3% ilaa 5% (3 ilaa 5 marka la soo qaado 100)
	Sickle Cell Disease	2% ilaa 30% (2 ilaa 30 marka la soo qaado 100)
Eeshiyada Bari iyo Koonfur-bari*	Alpha Thalassemia	5% (5 marka la soo qaado 100)
	Beta Thalassemia	2% ilaa 4% (2 ilaa 4 marka la soo qaado 100)
Hispanik*	Beta Thalassemia	0.25% ilaa 8% (in ka yar 1 ilaa 8 marka la soo qaado 100)
	Sickle Cell Disease	0.6% ilaa 14% (in ka yar 1 ilaa 14 marka la soo qaado 100)
Bariga Dhexe iyo Koonfur-dhexe Eeshiya*	Beta Thalassemia	0.5% ilaa 5.5% (in ka yar 1 ilaa 6 marka la soo qaado 100)
	Sickle Cell Disease	5% ilaa 25% (5 ilaa 25 marka la soo qaado 100)

** Lambarada kooxdan waa qiyaas waxaana laga yaaba in ay kala duwanaadaan iyadoo ay ku xidhan tahay asalka isirka.*

La talliyaha geneetikada wuxuu kugu kaalmeyn adiga iyo saaxiibkaa in aad gaartaan go'aano ku saabsan baarista uur jiifka.

Go'aan Qaadasshada in la Sameeyo Badhitaanadan

Doorasha in aad ka qaadan doonto wax uun baadhitaanadan, ama aad ogaato in mid ka ah u kuu wanaagsan yahay, oo adkaan karana. Doorasho "sax" ihii ma jirto. Dumarka qaarkood waxay doortaan oo kaliya badhitaanka ku saabsan qalfoofka jidhka oo kaliya. Kuwa kale waxa laga yaabaa in ay doortaan baadhitaan isudhafan iyo qalfoofka. Haddii baadhitaanka mid ka mid ihi u noqdo mid aan caadi ahayn waxay samayn karaan amniocentesis. Dumarka qaarkood waxay doortaan CVS ama amniocentesis iyaga oo aan samayn baadhitaanka screeningka.

Qaadashada Go'aan aan Rasmi Ahayn

Himiladena Caafimaadka UW waa in aan iskaashi la samayno bukaanka iyo qoysaska si go'aan looga gaaro daryeelkooda. Waxaan kugu dhiiri gelineynaa in aad weydiisid su'aalo kugu kaalmeeya in aad gaartid go'aano.

Kuwaan waa qaar ka mid ah su'aalaha laga yaabo in aad weydiisid naftaada inta aad ka fakareysid qaadashada baarista geneetikada:

- ❑ Miyaan u baahan ahay in aan helo macluumaadka oo dhan?
- ❑ Sidee bay barashada la xidhiidha ciladahan dhalasho inta aanu ilmahaygu dhalan u daryeelahayga caafimaad iigu diyaarinayaa iiguna qorshaynayaa?
- ❑ Sidee bay macluumaadkani iiga caawinayaa in aan doorto uurkayga haddii cilad la iga helo?
- ❑ Miyey qaadasha baadhitaanadani iga gacansiinaysaa in aan hubaal dareemo?

Daryeelahaaga caafimaad ayaa si faahfaahsan kaala hadli doona daryeekaaga caafimaad iyo wixii la xidhiidha doorashooyikaaga. Ama, waxaad isku qori doontaa kaltanka balamaha ee Kliinikada Baarista Uur Jiifka ka hor (Prenatal Diagnosis Clinic). La Talliyeyaasha geneetikada waxaa si gaar ah loo tababaray in ay dadka ku kaalmeyaan fakarka su'aalahaan. Waxay idinka caawinayaan si aad uga go'aan qaadataan waxa idiin wanaagsan.

Su'aalo?

Su'aalahaagu waa muhiim. Haddii aad qabtid su'aalo ku saabsan baarista uur jiifka, wac bixiyaha dareyaalka caafimaadkaada saacadaha xafiiska.

Waxaad kaloo wici kartaa Kliinikada Baarista Uur Jiifka: 206-598-4072

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Guide to Prenatal Testing

Learning about your baby's health

This chapter describes prenatal tests that give information about your baby's health. It is your choice whether or not to have these tests done. Talk with your health care provider to learn more and to help you decide if any of these tests are right for you.

If you have any of these tests done, you will be asked to read more about each one. You will also be asked to read and sign a consent form for each test.

There is a lot you can do during your pregnancy to keep you and your baby healthy. Taking prenatal vitamins, eating healthy foods, exercising, and getting enough sleep are all important.

The human body is complicated. Even if you do everything "right" during your pregnancy, babies do not always develop normally. Between 3% and 5% of babies (between 3 and 5 out of 100) have some kind of health problem when they are born.

This handout gives some basic information about these tests to help you make the best decision for you.

What are the tests?

There are 2 basic kinds of tests:

- **Screening tests** predict the *chance*, or odds, that your baby has a certain birth defect.
- **Diagnostic tests** tell you if your baby does or does not have a certain birth defect.

The tables on the next page list the tests and when they are done. They also give a brief description of each test and what it will tell you. The rest of this chapter gives more details about these tests, if you would like to read about them before you talk with your health care provider.

Prenatal tests can provide information about your baby's health.

Screening Tests

Name of Test	When	Description	What It Tells You
Nuchal translucency (NT) ultrasound	11 to 14 weeks	Abdominal ultrasound to measure small space behind baby's neck	*Chances* your baby has a chromosome problem
Integrated screen	11 to 14 weeks *and* 15 to 22 weeks	NT ultrasound plus 2 separate blood samples	*Chances* your baby has Down syndrome, trisomy 18, or spina bifida
Quad screen	15 to 22 weeks	1 blood sample	*Chances* your baby has Down syndrome, trisomy 18, or spina bifida

Diagnostic Tests

Name of Test	When	Description	What It Tells You
Chorionic villus sampling (CVS)	11 to 14 weeks	Sample of placenta, taken through the vagina or abdomen	*Whether or not* your baby has chromosome problems and sometimes other inherited diseases
Amniocentesis (with ultrasound)	16 to 22 weeks	Sample of fluid from around your baby, taken through your abdomen	*Whether or not* your baby has chromosome problems, spina bifida, and sometimes other inherited diseases

Other Tests

Name of Test	When	Description	What It Tells You
Anatomy ultrasound	18 to 22 weeks	Abdominal ultrasound to check baby's growth and development	*Whether or not* abnormalities are suspected and if further testing is needed

Screening Tests

Nuchal Translucency (NOO-kul trans-LOO-sun-see) or NT Ultrasound

This screening test is done between 11 and 14 weeks of pregnancy. Using ultrasound, your baby's length is measured to confirm your due date. Ultrasound is also used to measure the small space under the skin behind your baby's neck. This space is called the *nuchal translucency* (NT). The larger this space of fluid is, the greater the *chance* your baby has a chromosome problem. An NT ultrasound can be done only by specially trained staff.

Integrated (IN-tuh-grey-tud) screen

This test uses the results of the NT ultrasound and 2 blood tests. The first blood sample is taken between 11 and 14 weeks, usually the same day as the NT ultrasound. The 2nd blood sample is taken between 15 and 22 weeks. The blood tests look for patterns of proteins and hormones that are linked to certain birth defects.

An integrated screen tells you the *chances* that your baby has Down syndrome, trisomy 18, or spina bifida. (See the sidebars on pages 34, 35, and 36 for more details.) It does not diagnose these conditions. Most women who get an abnormal integrated screen result still have a healthy baby.

The integrated screen can detect:

- 90 out of 100 cases (90%) of Down syndrome
- 90 out of 100 cases (90%) of trisomy 18
- 80 out of 100 cases (80%) of spina bifida

But, it will **not** detect all cases of these birth defects. And, it does not test for any other health problems.

Quad Screen

This screening test involves 1 blood sample that is taken between 15 and 22 weeks. It's like the integrated screen, because it also looks for patterns of proteins and hormones that are linked to certain birth defects.

A quad screen tells you the *chances* that your baby has Down syndrome, trisomy 18, or spina bifida. It does not diagnose these conditions. Most women who get an abnormal quad screen result still have a healthy baby.

What is a chromosome problem?

Chromosomes are packages of genetic instructions. We inherit them from our parents. They control how our bodies grow and develop. They are in every part of our body, and they can only be seen with a microscope.

Most people have 46 chromosomes, but some people have an extra chromosome (47), a missing one (45), or a chromosome that has a piece broken off or an extra piece attached.

A change in a chromosome often causes birth defects and developmental delay, but not always.

The quad screen can detect:

- 85 out of 100 cases (85%) of Down syndrome
- 75 out of 100 cases (75%) of trisomy 18
- 80 out of 100 cases (80%) of spina bifida

But, it will **not** detect all cases of these birth defects. And, it does not test for any other health problems.

A quad screen may be a good test to have if you do not start prenatal care until your 4th month or if an NT ultrasound is not available.

Advanced Aneuploidy (ann-you-PLOY-dee) Screening with Cell-free DNA

You may have heard about a new blood test that can screen for Down syndrome. This test is called a*dvanced aneuploidy screening with cell-free DNA*. It uses a blood sample from the mother, and it is done starting at 10 weeks of pregnancy. It screens for specific chromosome disorders in the baby.

Everyone has some free (not contained within a cell) DNA in their blood. When you are pregnant, most of that cell-free DNA is from you, but some is from your pregnancy. In this test, the total amount of cell-free DNA from chromosomes 21, 18, and 13 is measured in your blood.

Like the other screening tests, this test does not tell you if the baby has, or does not have, a chromosome problem. But if there is an increased amount of DNA from one of these chromosomes in your blood, there is a high chance that the baby has trisomy for that chromosome.

Currently, only women who have a high risk of having a baby with Down syndrome, trisomy 18, or trisomy 13 can have this test. If you have already had a child with one of these trisomies, or if you have another type of screen and the results are abnormal, you may be offered advanced aneuploidy screening with cell-free DNA.

Diagnostic Tests

Anatomy (uh-NAT-uh-mee) Ultrasound

This test is done between 18 and 22 weeks. An ultrasound is used to look at your baby, the amount of fluid around him, your placenta, and your uterus. It checks to see that the baby is growing and that all major organs are formed.

What is Down syndrome?

Down syndrome is also known as trisomy 21. It is caused when a person has an extra copy of chromosome number 21.

Down syndrome affects people in different ways. People with Down syndrome always look different than other members of their family. They always have some developmental delay, but the level of delay differs from person to person.

Adults with Down syndrome may be able to play sports, have a basic job, and enjoy friends. But they usually cannot live on their own without help.

Many babies with Down syndrome have a heart defect, which can sometimes be fixed with surgery. Other health problems and birth defects sometimes occur with Down syndrome, but they are rare.

Your baby is developed enough at this age that an ultrasound may find problems such as a severe heart defect, spina bifida, a missing kidney, and severe cleft lip. Although this test will not diagnose chromosome problems, it may show signs of them or other conditions.

Chorionic Villus Sampling (kor-ee-ON-ic VILL-us sam-pling) or CVS

This diagnostic test is usually done between 11 and 14 weeks. The doctor uses either a thin, flexible needle or a thin plastic tube to remove a small sample of the placenta. An ultrasound is done at the same time, so your baby can be seen during the procedure.

The placenta sample is used to diagnose chromosome problems. If an inherited condition such as *muscular dystrophy* or *hemophilia* runs in your family, the sample can be used to test your baby for that condition.

The chance of miscarriage after CVS is 1 to 2 women in 100 (1% to 2%).

Amniocentesis (AM-nee-oh-sen-TEE-sis) or Amnio

This diagnostic test is usually done between 16 and 22 weeks. The doctor uses a thin, flexible needle to take 2 tablespoons of fluid from around your baby. An ultrasound is done at the same time, so your baby can be seen during the procedure.

The fluid is used to diagnose chromosome problems and spina bifida. If an inherited condition like muscular dystrophy or hemophilia runs in your family, the fluid can be used to test your baby for that condition.

The chance that having an amniocentesis will cause a miscarriage is 1 in 400 women (0.25%).

Ancestry-Based Carrier Screening

Your ancestry, or ethnicity, is one clue to help learn if your baby could have a rare genetic disease. Each ancestral group has conditions that can be inherited that are more common in that group compared to other ethnic groups. The conditions that are linked with each ancestral group are listed in the table on page 36.

Most times, a couple can have a child with one of these disorders only when *both* parents are "carriers" for the *same* disorder. **Carriers usually have no symptoms of the disease.** Also, most carriers have no family history of the disease. If someone in your family has one of these conditions, tell your health care provider.

What is trisomy 18?

Trisomy 18 is also known as Edwards syndrome. It occurs when a person has an extra copy of chromosome number 18.

Most babies with this condition do not survive the pregnancy. Children with trisomy 18 have severe brain damage and usually other problems, such as heart defects and clubfoot.

What is spina bifida?

Spina bifida is a condition in which part of the baby's spine does not form normally and the nerves in the spine are damaged. This happens within the first few weeks of pregnancy.

Spina bifida affects people in different ways. Some people have trouble walking and may need to use braces or a wheelchair. Some have trouble controlling their bladder or bowel. Sometimes, spina bifida can cause brain damage and developmental delay.

If you and your partner are both carriers for the same genetic condition, then your baby could inherit that condition. If you want to know for sure before birth, an amniocentesis or a CVS can be done. The integrated screen, quad screen, and ultrasound will **not** diagnose these disorders.

To see if you are a carrier for these hereditary conditions, you will need to give a small blood sample. It is your choice whether or not to have any or all of these tests.

This table is adapted from "Ancestry Based Carrier Screening," published by the National Society of Genetic Counselors, Inc., 2005:

Ancestral Group	Hereditary Condition	Chance of Being a Carrier
African-American	Beta Thalassemia	10% (10 out of 100)
	Sickle Cell Disease	11% (11 out of 100)
Eastern European (Ashkenazi) Jewish	Canavan Disease	2.5% (2 to 3 out of 100)
	Cystic Fibrosis	3% to 4% (3 to 4 out of 100)
	Familial Dysautonomia	3% (3 out of 100)
	Tay-Sachs Disease	3% (3 out of 100)
European Caucasian	Cystic Fibrosis	3% (3 out of 100)
Mediterranean	Beta Thalassemia	3% to 5% (3 to 5 out of 100)
	Sickle Cell Disease	2% to 30% (2 to 30 out of 100)
East and Southeast Asian*	Alpha Thalassemia	5% (5 out of 100)
	Beta Thalassemia	2% to 4% (2 to 4 out of 100)
Hispanic*	Beta Thalassemia	0.25% to 8% (fewer than 1 to 8 out of 100)
	Sickle Cell Disease	0.6% to 14% (fewer than 1 to 14 out of 100)
Middle Eastern and South Central Asian*	Beta Thalassemia	0.5% to 5.5% (fewer than 1 to 6 out of 100)
	Sickle Cell Disease	5% to 25% (5 to 25 out of 100)

** Numbers for this group are estimates and may vary depending on exact ethnicity.*

A genetic counselor can help you and your partner make decisions about prenatal tests.

Deciding Whether to Do These Tests

Choosing whether to have any of these tests, or deciding which ones are best for you, can be hard. There is no "right" choice. Some women choose only an anatomy ultrasound and no other tests. Others may choose an integrated screen and anatomy ultrasound. And, if one of these tests is abnormal, they may have amniocentesis. Some women prefer a CVS or amniocentesis without any of the screening tests.

Making an Informed Decision

Our goal at UW Medicine is to partner with patients and families in making decisions about their care. We encourage you to ask questions to help you to make your decisions.

These are some questions you may want to ask yourself as you think about having genetic testing:

- ❑ Do I want to have any of this information?
- ❑ How would learning about these birth defects before my baby is born help me and my health care provider prepare and plan?
- ❑ How would this information help me make choices about my pregnancy if a birth defect is found?
- ❑ Will taking these tests help me feel more reassured?

Your health care provider can talk more with you about your choices. Or, you can schedule an appointment in the Prenatal Diagnosis Clinic. Genetic counselors are specially trained to help people think through these questions. They can help you make the decision that is best for you.

Questions?

Your questions are important. If you have questions about prenatal testing, call your health care provider during office hours.

You may also call the Prenatal Diagnosis Clinic: 206-598-4072

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