RARE resources WALES

2 Y daith at ddiagnosis

2 The journey to diagnosis

Mae Rare Resources yn gasgliad o ganllawiau gwybodaeth i deuluoedd sydd wedi cael diagnosis o gyflwr genetig neu brin yn ddiweddar, sydd ar y daith at ddiagnosis, neu sydd wedi cael gwybod bod cyflwr eu plentyn mor brin fel na fyddant yn cael diagnosis efallai.

Datblygwyd y canllawiau Rare Resources mewn cydweithrediad rhwng Genetic Alliance UK a theuluoedd yng Nghymru. Mae'r canllawiau'n rhoi dolenni at ffynonellau gwybodaeth a chymorth dibynadwy, ac yn cynnwys 'awgrymiadau da' gan deuluoedd eraill.

Mae Rare Resources yn cynnwys y canllawiau canlynol:
1. Eglurhad o gyflyrau genetig, prin a heb ddiagnosis
2. Y daith at ddiagnosis
3. Defnyddio'r GIG yng Nghymru
4. Cymorth a gwybodaeth i rieni a gofalwyr
5. Cymorth a gwybodaeth i'ch plentyn
6. Cyfeiriadur gwybodaeth sy'n rhoi manylion y gwasanaethau cymorth sydd ar gael yng Nghymru

Gellir lawrlwytho'r canllawiau Rare Resources o bit.ly/rareresourceswales

Rare Resources is a collection of information guides for families who have recently received a diagnosis of a genetic or rare condition, are on the journey to a diagnosis, or have been told their child's condition is so rare they might not get a diagnosis.

The Rare Resources guides have been developed in collaboration between Genetic Alliance UK and families in Wales. The guides provide links to reliable sources of information and support, and contain 'top tips' from other families.

Rare Resources contains the following guides:
1. Genetic, rare and undiagnosed conditions explained
2. The journey to diagnosis
3. Using the NHS in Wales
4. Support and information for parents and carers
5. Support and information for your child
6. Information directory detailing support services available in Wales

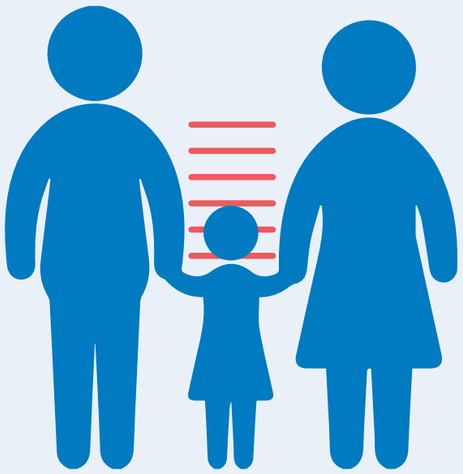
The Rare Resources guides can be downloaded from bit.ly/rareresourceswales

GENETIC ALLIANCE UK

RARE resources WALES

2.1 Beth i'w wneud os ydych chi'n poeni am ddatblygiad eich plentyn

2.1 What to do if you are worried about your child's development?



Os ydych chi'n poeni am ddatblygiad eich plentyn, mae'n syniad da trafod hyn efo eich Ymwelydd Iechyd neu Feddyg Teulu.

If you have concerns about your child's development, it is a good idea to discuss this with your Health Visitor or GP.

Dylai eich Ymwelydd Iechyd neu Feddyg Teulu wrando ac yna trafod eich pryderon efo chi, archwilio eich plentyn ac ystyried opsiynau. Os nad ydynt yn gallu cynnig diagnosis, gallant eich cyfeirio at weithwyr iechyd proffesiynol eraill am eu barn.

Gall eich meddyg teulu gyfeirio eich plentyn at baediatregydd am eu barn. Meddyg yw paediatregydd sydd wedi cael hyfforddiant arbennig ar ofal meddygol i fabis a phlant.

Os yw eich meddyg teulu neu baediatregydd yn amau cyflwr neilltuol, neu fath neilltuol o gyflwr, efallai y caiff eich plentyn ei anfon at glinigydd arbenigol.

Gall eich meddyg teulu hefyd eich cyfeirio at nyrs neu weithiwr gofal iechyd proffesiynol all helpu efo rhai symptomau. Er enghraifft, gall plentyn sydd â gwendid yn ei gyhyrau gael ei gyfeirio at ffisiotherapydd. Efallai bydd nyrsys a gweithwyr iechyd proffesiynol yn gallu eich helpu i gael diagnosis.

Os bydd eich meddyg teulu, paediatregydd neu glinigydd arbenigol yn amau cyflwr genetig, byddant yn trefnu i chi gael eich cyfeirio at Wasanaeth Geneteg y GIG. Gallwch chithau hefyd ofyn am gael eich cyfeirio.

Gall gymryd amser hir i gael diagnosis, ac efallai na fydd diagnosis yn bosibl i rai teuluoedd. Er hynny, mae cymorth ar gael i deuluoedd ar bob cam o'u taith.

Your Health Visitor or GP should listen and then discuss your concerns with you, examine your child and consider options. If they are unable to offer a diagnosis, they may refer you to other health professionals for their opinion.

Your GP may refer your child to a paediatrician for their opinion. A paediatrician is a doctor who has special training in medical care for babies and children.

If your GP or paediatrician suspects a particular condition, or a particular type of condition, your child may be referred to a specialist clinician.

Your GP may also refer you to a nurse or a healthcare professional who can help with particular symptoms. For example, a child experiencing muscle weakness may be referred to a physiotherapist. Nurses and health professionals may be able to help you obtain a diagnosis.

If a genetic condition is suspected by your GP, paediatrician or specialist clinician, a referral to an NHS Genetics Service will be arranged for you. You can also ask for this referral to be made.

It may take a long time to find a diagnosis and for some families, a diagnosis may not be possible. However, there is support available to families at all stages of their journey.

2.2 Sut i gael profion genetig i'ch plentyn

2.2 How to access genetic testing for your child

Er mwyn cael profion genetig, rhaid i chi gysylltu â'ch meddyg teulu neu glinigydd arbenigol a fydd yn trefnu i chi gael eich cyfeirio at Wasanaeth Geneteg Rhanbarthol y GIG.

To access genetic testing you must contact your GP or specialist clinician who will arrange a referral for you to an NHS Regional Genetics Service.

Yma byddwn yn egluro beth fydd yn digwydd yn yr apwyntiad hwnnw a pha mor hir y gallech chi orfod aros am ganlyniad.

Mae profion genetig ar y GIG ar gael drwy eich clinigydd arbenigol sy'n gofyn am y profion sy'n briodol i chi o labordy GIG ardystiedig.

Bydd y clinigydd yn gofyn am brawf dim ond os yw'n gwybod y bydd y canlyniadau yn ei helpu i roi'r gofal iechyd mwyaf priodol i chi. Mae polisïau'r GIG yn diffinio pwy sydd fwyaf tebygol o elwa o gael profion genetig penodol. Bydd eich clinigydd arbenigol yn casglu eich sampl am ddadansoddiad genetig a'i anfon i'r labordy. Yna bydd y labordy yn dadansoddi a dehongli'r canlyniadau.

Wedyn, bydd eich clinigydd ar gael i fynd drwy'r canlyniadau efo chi a beth maent yn ei olygu. Mae unrhyw un sy'n cael prawf genetig ar y GIG hefyd yn debygol o weld cwnselydd genetig.

Os bydd y prawf yn edrych i weld a oes gennych gyflwr genetig difrifol, bydd cwnsela ar gael cyn i chi gael y prawf ac wedyn.

Here we explain what will happen at that appointment, and how long you might have to wait for a result.

Genetic testing on the NHS is available through your specialist clinician who orders the tests that are appropriate for you from an NHS-certified laboratory.

The clinician will request a test only if they know that the results will help them provide you with the most appropriate healthcare. NHS policies define who is most likely to benefit from specific genetic tests. Your specialist clinician will collect your sample for genetic analysis and send it to the laboratory. The laboratory will then analyse and interpret the results.

Your clinician will then be available to talk you through what your results mean. Anyone having a genetic test on the NHS is also likely to see a genetic counsellor.

If the test will be looking to determine whether you are affected by a serious genetic condition, counselling will be available both before and after you take the test.

2.3 Beth yw cwnsela genetig?

2.3 What is genetic counselling?

Mae cwnsela genetig yn wasanaeth sy'n darparu cymorth, gwybodaeth a chyngor am gyflyrau genetig.

Genetic counselling is a service that provides support, information and advice about genetic conditions.

Gweithwyr gofal iechyd proffesiynol sydd wedi cael hyfforddiant ar wyddor geneteg ddynol (cwnselydd genetig neu genetegydd clinigol) sydd yn rhoi'r cwnsela. Mae'r hyn a fydd yn digwydd yn ystod cwnsela genetig yn dibynnu pam yn union rydych chi wedi cael eich cyfeirio.

Gall olygu:

- dysgu am gyflwr iechyd sydd yn rhedeg yn eich teulu, sut mae'n cael ei etifeddu, a pha aelodau o'r teulu allai'r cyflwr effeithio arnynt
- asesiad o'r risg i chi a'ch partner basio cyflwr etifeddol ymlaen i'ch plentyn
- edrych ar hanes meddygol eich teulu neu deulu eich partner a llunio coeden deulu
- cymorth a chyngor os oes gennych blentyn sydd â chyflwr etifeddol a'ch bod chi eisiau plentyn arall
- trafodaeth am brofion genetig y gellir eu trefnu os yw hynny'n briodol, gan gynnwys risgiau, manteision a chyfyngiadau profion genetig
- help i ddeall canlyniadau profion genetig a beth maent yn ei olygu
- gwybodaeth am grwpiau cymorth i gleifion sy'n berthnasol
- Fe gewch wybodaeth glir a manwl gywir er mwyn i chi allu penderfynu beth sydd orau i chi.

Fel arfer, bydd eich apwyntiad yn digwydd yn eich canolfan geneteg GIG agosaf.

Mwy o wybodaeth:
https://bsgm.org.uk/public-patients-families

It's conducted by healthcare professionals who've received training in the science of human genetics (a genetic counsellor or a clinical geneticist). What happens during genetic counselling will depend on exactly why you've been referred.

It may involve:

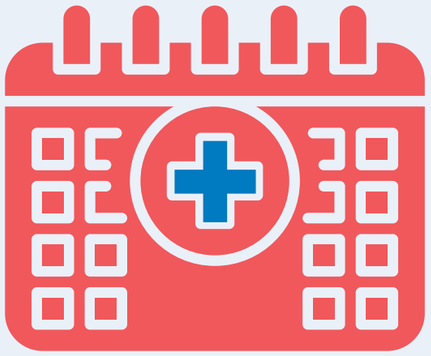
- learning about a health condition that runs in your family, how it's inherited, and which family members may be affected
- an assessment of the risk of you and your partner passing an inherited condition on to your child
- a look at the medical history of your family or your partner's family and drawing up a family tree
- support and advice if you have a child affected by an inherited condition and you want to have another child
- a discussion about genetic tests, which can be arranged if appropriate, including the risks, benefits and limitations of genetic testing
- help understanding the results of genetic tests and what they mean
- information about relevant patient support groups
- You'll be given clear, accurate information so you can decide what's best for you.

Your appointment will usually take place at your nearest NHS genetics centre.

Further Information:
https://bsgm.org.uk/public-patients-families

2.4 Beth sy'n digwydd mewn apwyntiad profi genetig?

2.4 What happens at a genetic testing appointment?



Ar ôl cael eich cyfeirio at Wasanaeth Geneteg Rhanbarthol y GIG, anfonir taflen neu lythyr atoch fel arfer yn egluro beth fydd yn digwydd pan fyddwch chi'n mynd i'r clinig am eich apwyntiad. Mae'r llythyr hwn yn aml yn gofyn i chi ddod â'ch plentyn efo chi i'r apwyntiad.

After being referred to an NHS Regional Genetics Service you will usually be sent a leaflet or letter explaining what will happen when you attend the clinic for your appointment. This letter often asks you to bring your child with you to the appointment.

Cyn mynd i apwyntiadau, mae'n syniad da nodi unrhyw gwestiynau rydych chi eisiau eu gofyn pan fyddwch chi yno.

Efallai y byddai'n syniad i chi fynd â llyfr nodiadau efo chi i'r apwyntiad hefyd. Mae rhai teuluoedd yn recordio'r apwyntiad ar eu ffôn er mwyn iddynt allu gwrando eto wedyn. Dylech wastad ofyn am ganiatâd pobl eraill sydd yn yr apwyntiad cyn recordio.

Os ydych chi'n poeni na fyddwch chi'n cofio rhai o'r manylion yn yr apwyntiad, gallwch ofyn i'ch genetegydd eu hysgrifennu nhw i chi edrych arnynt ato pan ewch chi adref.

Fel arfer, byddant yn ysgrifennu atoch ar ôl yr apwyntiad yn crynhoi'r hyn a drafodwyd fel bod gennych gofnod ysgrifenedig ohono.

Os ydych chi'n rhiant sengl neu fod eich partner methu dod i'r apwyntiad, yna efallai y byddwch chi eisiau dod ag aelod arall o'r teulu efo chi neu ffrind dibynadwy am gymorth. Efallai y byddant yn gallu cofio pethau am yr apwyntiad y byddwch chi wedi'u methu.

Yn aml ar ddechrau'r apwyntiad cyntaf, bydd y genetegydd yn gofyn i rieni beth maent yn gobeithio cael gwybod, er enghraifft:

- Diagnosis i'w plentyn
- Beth fydd yr effaith ar eu plentyn yn y dyfodol
- Allai hyn effeithio ar unrhyw blant a gânt yn y dyfodol
- A allai plant eraill yn y teulu ehangach fod wedi'u heffeithio

Wedi hynny, bydd y rhan fwyaf o'r apwyntiad yn cael ei dreulio yn mynd drwy'r 'broses asesu genetig.'

Proses ddiagnostig yw hyn ble mae genetegwyr clinigol yn gweithio gyda staff labordy a gweithwyr meddygol proffesiynol eraill i edrych ar y rhesymau posibl am y patrymau datblygiadol gwahanol sydd gan blentyn.

Maent yn gobeithio adnabod syndrom neu gyflwr genetig sy'n egluro anawsterau'r plentyn. Efallai mai anhwylder un cromosom yw achos y cyflwr, neu efallai

Before attending appointments, it is a good idea to write down any questions that you want to ask when you are there.

You might also find it helpful to take a notebook along to the appointment. Some families record the appointment on their phone so they can listen again afterwards. You should always ask permission from the other people at the appointment before recording it.

If you are worried you won't remember some of the details at the appointment you can ask your geneticist to write them down for you to look at again when you get home.

Usually they will write to you after the appointment summarising what was discussed so you have a written record of it.

If you are a single parent or your partner cannot attend the appointment you might want to take along another family member or trusted friend for support. They may also remember points about the appointment that you miss.

Often at the start of the first appointment the geneticist will ask parents what they are hoping to find out, for example:

- A diagnosis for their child
- How their child will be affected in the future
- If any future children could be affected
- Whether other children in the wider family could be affected

Most of the appointment will then be going through the 'genetic assessment process'.

This is a diagnostic process where clinical geneticists work with laboratory staff and other medical professionals to explore the possible reasons for the different developmental patterns a child has.

Their aim is to identify a genetic syndrome or condition that explains the child's difficulties. The cause of the condition might be a single-gene disorder, or it might be that a particular chromosome has a piece missing (deletion) or an extra piece (duplication) or it may be

bod darn ar goll (dileu) neu ddarn ychwanegol (dyblygu) mewn cromosom neilltuol, neu efallai bod darnau o gromosomau wedi ‘ffeirio lle’ (trawsleoli).

Mae adnabod achos genetig problemau iechyd plentyn yn digwydd drwy gyfuniad o ymchwiliadau:
- Holi’n fanwl am ddatblygiad y plentyn
- Holi am hanes meddygol teulu’r plentyn
- Archwiliad corfforol gan gynnwys lluniau
- Ymchwiliadau labordy (os bydd angen, caiff samplau gwaed eu tynnu yn ystod yr apwyntiad neu’n fuan wedyn)

Holi’n fanwl am ddatblygiad y plentyn

Mae hanes datblygiad plentyn a phatrwm ei broblemau ar hyn o bryd, neu o’r blaen, yn elfen allweddol o’r asesiad. Gall fod yn rhwystredig gorfod ailadrodd hanes eich plentyn i weithiwr proffesiynol arall eto, ond mae hyn yn rhan bwysig iawn o’r asesiad.

Gall hyn fod yn dorcalonnus weithiau oherwydd bod rhaid i chi ganolbwyntio ar yr holl bethau sy’n gwneud eich plentyn yn wahanol neu bethau nad yw’n gallu eu gwneud.

Mae’n syniad da siarad efo rhywun ar ôl eich apwyntiad.

Gall fod yn ffrind neu’n aelod o’r teulu, neu efallai eich bod chi eisiau rhannu eich teimladau efo teuluoedd eraill sy’n deall pa mor anodd all yr apwyntiadau hyn fod.

Holi am hanes meddygol teulu’r plentyn

Bydd y genetegydd hefyd yn holi am hanes eich teulu i weld a oes patrymau allai roi cliwiau am achos problem eich plentyn.

Mae hanes teulu hefyd yn bwysig oherwydd os yw’r broblem yn un genetig, yna gall hyn fod oherwydd newidiadau mewn genynnau neu gromosomau gafodd eu pasio i lawr gan un rhiant, neu’r ddau riant. Weithiau, bydd hi’n bosibl gwneud diagnosis penodol ar sail y disgrifiadau hanesyddol hyn yn unig, heb fod angen profion genetig.

Weithiau, gall fod yn anodd i deuluoedd ddisgrifio hanes eu teulu oherwydd eu bod wedi cael eu mabwysiadu neu heb gael fawr o gyswllt â’u teulu.

Archwiliad corfforol gan gynnwys lluniau

Bwriad hyn yw adnabod unrhyw nodweddion corfforol allai roi cliw i’r genetegydd am ddiagnosis posibl.

Gall hyn fod yn broses ryfedd ac annifyr weithiau, i’r plentyn a’r rhieni.

that pieces of chromosomes have ‘swapped around’ (translocation).

Identifying the genetic cause of a child’s health problems occurs through a combination of investigations:
- Asking about the child’s development in detail
- Asking about the child’s family’s medical history
- Physical examination including photographs
- Laboratory investigations (if necessary blood samples are taken are taken during the appointment or shortly afterwards)

Asking about the child’s development in detail

The history of a child’s development and the pattern of problems that they currently have or have had in the past is a key component of the assessment. It can be frustrating having to repeat your child’s history to yet another professional, but this is a very important part of the assessment.

This can sometimes feel upsetting as you have to focus on all the things that make your child different or that they can’t do.

It is a good idea to have people you can talk to after your appointment.

This might be a friend or family member or you might want to share your feelings with other families who understand how difficult these appointments can be.

Asking about the child’s family’s medical history

The geneticist will also ask about your family history to see if they can identify patterns that might give clues to the cause of your child’s problem.

Family history is also important because if the problem is genetic then it may be the result of changes in genes or chromosomes which were passed down from one or both of the parents. Sometimes it may be possible to make a specific diagnosis based on these historical descriptions alone without the need for genetic testing.

Sometimes families might find it difficult to describe their family histories because they may be adopted or have little contact with their family.

Physical examination including photographs

The aim of this is to identify any particular physical features that might give the geneticist a clue about a possible diagnosis.

This can be an odd, and sometimes upsetting, process for both the child and parents.

Ymchwiliad labordy (os bydd angen, cymerir samplau gwaed yn ystod yr apwyntiad neu’n fuan wedyn).

Dyma ran olaf y broses ddiagnostig fel arfer. Mae’n digwydd ar ôl astudio hanes teulu’r plentyn yn fanwl a’r archwiliad corfforol.

Bydd samplau DNA y plentyn (a’r rhieni weithiau) yn cael eu cymryd i’w profi ac edrych am newidiadau yn y DNA. Mae’r samplau sydd eu hangen yn gallu amrywio a gallant gynnwys gwaed, salifa neu groen.

Unwaith y bydd gan genetegydd syniad am ddiagnosis posibl i’r plentyn, gall ofyn am brofion labordy ar unwaith i gadarnhau’r diagnosis y mae’n ei amau.

Bydd rhaid aros yn hir am ganlyniadau yn aml.

Mae’n syniad siarad â rhywun ar ôl eich apwyntiad profi genetig.

Pa mor hir mae’n ei gymryd i gael canlyniadau profion genetig?

Weithiau nid yw’n bosib gwneud profion genetig ar unwaith. Efallai bod eich plentyn yn rhy ifanc er mwyn i’w nodweddion corfforol roi unrhyw ‘gliw’ clir am beth allai’r cyflwr fod.

Yn y sefyllfaoedd hyn, byddwch chi fel arfer yn cael eich gwahodd yn ôl am apwyntiadau dilynol rai blynyddoedd wedyn pan fydd mwy o gliwiau oherwydd bod eich plentyn wedi datblygu, bod gwybodaeth feddygol newydd wedi dod i’r amlwg, neu fod prawf newydd ar gael. ‘Aros gwyliadwrus’ y mae genetegwyr fel arfer yn galw’r cyfnod hwn.

Mae’r cyfnod hwn, sef ‘aros gwyliadwrus’ yn synnu teuluoedd; nid yw’r rhan fwyaf o bobl yn sylweddoli pa mor hir all profi genetig ei gymryd.

Gall fod yn rhwystredig ac yn boenus gorfod aros am atebion a bydd llawer o deuluoedd yn meddwl tybed a ydynt wedi mynd yn angof. Tra byddant yn aros, bydd llawer o deuluoedd yn dechrau edrych ar y rhyngrwyd i chwilio am atebion ynghylch beth sy’n bod efo’u plentyn.

Er bod hyn yn naturiol, mae chwilio ar y we am symptomau yn aml yn gallu cynhyrchu lluniau o blant sydd â llawer o wahanol gyflyrau all amrywio yn y ffordd y maent yn effeithio ar wahanol blant.

Yn aml, lluniau o’r sefyllfa waethaf sy’n dod i fyny ar y sgrin ac mae rhai o’r delweddau hyn yn gallu bod yn dorcalonnus iawn i deuluoedd eu gweld.

Mae profi genetig yn gallu cymryd amser hir ac efallai bydd rhaid i chi aros am rai blynyddoedd cyn cael unrhyw ganlyniad.

Laboratory investigations (if necessary blood samples are taken during the appointment or shortly afterwards)

This is usually the final part of the diagnostic process. It takes place after careful examination of the child’s history and the physical examination.

Samples of the child’s DNA (and sometimes the parents’) will be taken to test and look for changes in the DNA. The samples needed can vary and may include blood, saliva or skin.

Once a geneticist has an idea about what might be a potential diagnosis for the child, they may order laboratory tests straight away to confirm the suspected diagnosis.

There can often be a long wait for results.

It is helpful to have people you can talk to after your genetic testing appointment.

How long does it take to get results from genetic testing?

Sometimes it is not possible to undertake genetic testing straight away. This might be because your child is too young for their physical features to give any clear ‘clue’ about what the underlying condition may be.

In these situations, you will usually be invited back for follow-up appointments several years later when there may be more clues because your child has developed, because new medical knowledge has come to light, or because a new test has become available. This period is usually referred to by geneticists as ‘watchful waiting’.

It is common for families to be surprised by the ‘watchful waiting’ period; most people don’t realise how long genetic testing can take.

It can be frustrating and worrying waiting for answers and many families wonder if they have been forgotten. Whilst they are waiting, many families start searching the internet to see if they can find answers about what is wrong with their child.

Although this is understandable, internet searches for symptoms often produce photographs of children with a range of different conditions which can vary in the way they affect different children.

It is often the ‘worst-case scenario’ images that pop up and some of these images can be very upsetting for families to see.

Genetic testing can take a long time and you may have to wait several years for any result.

2.5 Paratoi am apwyntiadau

2.5 Preparing for appointments

Efallai bydd rhaid i chi fynd i nifer o apwyntiadau, efo nifer o wahanol weithwyr gofal iechyd proffesiynol ar hyd eich taith at gael diagnosis i'ch plentyn.

You may have to attend a number of appointments, with a number of different health professionals on your journey to a diagnosis for your child.

Mae'n naturiol i chi deimlo dan straen ac yn bryderus am fynd i apwyntiadau – mae teuluoedd yn dweud wrthym bod paratoi am apwyntiadau yn gallu helpu efo'r teimladau hyn weithiau.

Mae'n syniad da paratoi rhestr o bethau yr hoffech chi eu trafod yn ystod eich apwyntiad.

Gall y rhain fod yn bethau yr ydych chi eisiau dweud wrth eich meddyg teulu neu glinigydd arbenigol – er enghraifft, symptomau newydd sydd gan eich plentyn neu unrhyw newidiadau neu batrymau ymddygiad.

Bydd rhai teuluoedd yn gweld ei bod hi'n help gwneud fideos i fynd efo nhw i ddangos i glinigydd arbenigol eu plentyn, er enghraifft, os ydych chi'n meddwl bod eich plentyn yn cael ffit (*seizure*) neu'n ymddwyn mewn ffyrdd anarferol.

Mae'n syniad da hefyd nodi unrhyw gwestiynau yr hoffech chi eu gofyn pan fyddwch chi yno.

Gall fod yn help mynd â phartner, aelod teulu, ffrind neu eiriolwr efo chi i'ch apwyntiad. Gallant gynnig cymorth, ond hefyd eich atgoffa chi i ofyn unrhyw gwestiynau yr oeddech chi eisiau eu gofyn ac efallai y byddant yn cofio rhywbeth ddywedwyd yn y cyfarfod 'ddaru chi ei fethu.

Mae'n syniad da cadw manylion eich apwyntiadau, llythyrau a chanlyniadau profion efo'ch gilydd. Fel hyn, gallwch gadw cofnod o daith eich plentyn tuag at ddiagnosis.

It is normal to feel overwhelmed and anxious about attending appointments – families tell us that preparing for appointments can sometimes help with these feelings.

It is a good idea to prepare a list of things you would like to discuss during your appointment.

This can be things you want to tell your GP or specialist clinician –for example, new symptoms your child has had or any changes or patterns of behaviour.

Many families find that it is helpful to make videos to take along to show their child's specialist clinician, for example, if you think your child might be having a seizure or is displaying unusual behaviours.

It's also a good idea to write down any questions that you would like to ask when you are there.

You may find it helpful to take a partner, family member, friend or advocate to your appointment. They can offer support, but also remind you to ask any questions you wanted to ask and they might remember something from the meeting that you missed.

It is a good idea to keep details of your appointments, letters and test results together. This will allow you to keep a record of your child's journey to diagnosis.

2.6 Beth mae cael diagnosis yn ei feddwl?

2.6 What does getting a diagnosis mean?

I'r rhan fwyaf o deuluoedd, mae cael diagnosis yr un mor bwysig wrth i'w plentyn fynd yn hŷn. Heb ddiagnosis, gall teuluoedd weld ei bod hi'n anodd cael gafael ar y cymorth iawn.

For most families getting a diagnosis remains just as important to them as their child grows up. Without one, families can struggle to access the right support.

Nid oes ganddynt unrhyw syniad sut ddyfodol fydd gan eu plentyn, neu a allai unrhyw blant eraill a gânt yn y dyfodol gael yr un cyflwr.

'Nid yw'r dymuniad i gael diagnosis yn golygu disgwyl am wellhad neu ffon hud ... ond mae gwybodaeth yn bwerus.'
Aelod SWAN UK

Er bod rhieni yn gyffredinol yn gwybod na fydd cael diagnosis yn newid bywyd eu plentyn yn sylweddol, maent yn gwybod y bydd yn rhoi mwy o synnwyr iddynt o'r hyn allant ei ddisgwyl.

Mae teuluoedd plant sydd wedi cael diagnosis genetig o gyflwr cromosom prin iawn, ble all y plentyn fod yn un o'r ychydig rai yn y byd sydd â'r cyflwr hwnnw, yn gallu teimlo'n siomedig weithiau gyda'r wybodaeth a gânt.

Ar y cyfan, mae teuluoedd yn gobeithio y bydd diagnosis yn rhoi syniad iddynt sut ddyfodol fydd gan eu plentyn, ond nid felly mo'r achos pob tro.

Er bod teuluoedd fel arfer yn falch eu bod nhw bellach yn gallu egluro beth sy'n bod efo'u plentyn, nid yw'r disgrifiad technegol o'r newid genetig yng ngenynnau neu gromosomau eu plentyn yn rhoi fawr o syniad iddynt o'r hyn allant ei ddisgwyl yn y dyfodol. Os mai eu plentyn nhw yw'r unig un, neu'r un o'r ychydig rai yn y byd sydd â'r newid genetig hwn, efallai y byddant yn dal i deimlo'n eithaf unig.

'Doeddwn i ddim yn disgwyl gwellhad, roeddwn i'n gwybod bod cyflwr genetig yn un am oes, ond roeddwn i'n disgwyl cael deall yr hyn roedd o'n ei olygu i fywyd fy mab. Yr hyn a gefais i oedd llythyrau di-rif a rhifau'.
Aelod SWAN UK

Er hynny, er nad yw diagnosis prin wastad yn dweud rhyw lawer wrth y teulu am yr hyn allant ei ddisgwyl yn nyfodol eu plentyn, mae'n rhoi rheswm iddynt o leiaf am gyflwr eu plentyn – hyd yn oed os nad yw llawer o bobl tu allan i'r maes geneteg yn debygol o'i ddeall.

They have no idea what the future holds for their child or if other children they may have in the future could be affected.

'Wanting a diagnosis is not about expecting a cure or a magic wand ... but knowledge is power.'
SWAN UK member

Although parents generally know that having a diagnosis will not significantly change their child's life, they hope it will give them a greater sense of what to expect.

Families of children who have been given a genetic diagnosis of a very rare chromosome condition, where the child may be one of only a few in the world known with that condition, can sometimes feel disappointed with the information given to them.

Families generally hope that a diagnosis will give them an indication as to what the future holds for their child but this is not always the case.

Although families are usually glad they can now explain what is wrong with their child, the technical description of the genetic change in their child's genes or chromosomes does not give them much idea of what to expect from the future. If their child is the only one, or one of a few in the world with this known genetic change, they can still feel quite isolated.

'I wasn't expecting a cure, I knew a genetic condition was lifelong, but I was expecting an understanding of what it meant for my son's life. What I got was a string of letters and numbers.'
SWAN UK member

However, while these rare diagnoses do not always tell the family much about what they can expect in their child's future, it does at least give them a reason for their child's condition – even if it is unlikely to be understood by many people outside of the field of genetics.

‘Rydw i’n aml yn cael fy nal rhwng peidio â gadael i ddiagnosis H ddiffinio ei bywyd ac, ar yr un pryd, codi ymwybyddiaeth o gyflyrau prin a’r hyn maent yn ei olygu i deuluoedd fel un ni. Rydw i’n aml yn dweud y byddwn i’n dymuno cael gwared ar heriau H ond na fyddwn i’n tynnu ei genynnau ‘ciami’ oddi arni. Maen nhw’n gymaint o ran ohoni – byddai eu newid nhw yn ei newid hithau, y ferch rydym ni’n ei charu, yn gyfan gwbl mae’n debyg.’
Rhiant plentyn sydd ag Inv Dup Del 8P

Heb ddiagnosis, mae llawer o deuluoedd yn dweud eu bod nhw’n teimlo eu bod nhw’n cael eu trin fel ‘mam niwrotig’ neu fod pobl yn dweud wrthynt y bydd eu plentyn yn ‘dal fyny’ neu’n ‘tyfu allan ohono’. Drwy gael diagnosis, gallant ymateb i’r math hyn o sylwadau.

Mae cael diagnosis yn hynod o bwysig i deuluoedd, yn ymarferol ac yn emosiynol.

Dyma rai o’r prif resymau pam fod angen diagnosis:
- Cael syniad o sut fydd y dyfodol
- Heb ddiagnosis, nid oes gan deuluoedd syniad o sut fydd y dyfodol i’w plentyn – fydd o neu hi yn cerdded? Yn siarad? Yn cael disgwyliad oes byrrach?
- Gallai cael diagnosis helpu i adnabod triniaethau neu broblemau iechyd posibl y bydd angen cadw llygad arnynt yn y dyfodol. Heb ddiagnosis, mae’n anodd gwybod a yw symptomau newydd yn rhywbeth i boeni amdanynt neu beidio.
- Cael pobl i gymryd anghenion y plentyn neu’r person ifanc o ddifri.
- Gwybod a fydd effaith ar blant eraill yn y teulu – heb ddiagnosis, mae’n amhosibl gwybod ai wedi’i etifeddu y mae’r cyflwr neu wedi digwydd unwaith yn unig. Mae hyn yn golygu nad yw teuluoedd yn gwybod a fydd yr un cyflwr anhysbys yn effeithio ar unrhyw blant yn y dyfodol. Mae hyn yn effeithio ar rieni’r plentyn heb ddiagnosis, ond hefyd ei frodyr a’i chwiorydd, modrybedd, ewythrod a chefndryd.
- Cael gwasanaethau a chymorth: heb ddiagnosis, mae teuluoedd yn aml yn ei chael hi’n anodd mynd at wasanaethau. Yn aml ar ffurflenni neu feini prawf asesu, mae bocs sy’n gofyn am ddiagnosis y plentyn ac mae teuluoedd yn cael trafferth gwybod beth i’w ysgrifennu.

‘I ni, mae cael diagnosis yn golygu y gallwn gysylltu â theuluoedd a rhannu ein bywyd, y llon a’r lleddf, dathliadau ac amseroedd anodd efo eraill sy’n mynd ar hyd llwybr tebyg.’
Rhiant plentyn sydd ag Inv Dup Del 8P

‘I am regularly caught between not letting H’s diagnosis define her life and also raising awareness of rare conditions and what they mean for families like ours. I often say that although I wish I could take away H’s challenges I wouldn’t take away her wonky genes. They are so intrinsically her that to change them would probably completely change her, the person we love.’
Parent of a child with Inv Dup Del 8P

Many families say that without a diagnosis they feel like they are treated like a ‘neurotic parent’ or told that their child will ‘catch up’ or ‘grow out of it’. Having a diagnosis gives them a way to respond to this kind of comment.

Having a diagnosis is really important to families on a practical and emotional level.

Here are some of the main reasons for needing a diagnosis:
- To have a sense of what the future holds
- Without a diagnosis, families have no idea what the future holds for their child. Will they walk? Will they talk? Will they have a shorter life expectancy?
- Having a diagnosis could help identify potential treatments or health issues that need to be monitored in future. Without a diagnosis it is hard to know whether new symptoms are something to be worried about.
- To have the child or young adult’s needs taken seriously
- To know if other children in the family will be affected – without a diagnosis it is impossible to know if the condition is inherited or just a one off. This means families don’t know if future children will be affected by the same unknown condition. This affects not only the parents of the undiagnosed child, but also their siblings, aunts, uncles and cousins.
- To access service and support: families often find it hard to access services without a diagnosis. Forms or assessment criteria often have a box that asks for the child’s diagnosis and families struggle to know what to write.

‘For us having a diagnosis means we can connect with families and share our lives, ups and downs, celebrations and hard times with others walking a similar path.’
Parent of a child with Inv Dup Del 8P

2.7 Pam ei bod hi'n anodd gwneud diagnosis o rai cyflyrau?

2.7 Why are some conditions difficult to diagnose?

Mae'r daith hir ac anodd sy'n wynebu llawer o deuluoedd er mwyn cael diagnosis sydd wedi'i gadarnhau, yn aml yn cael ei alw'n daith ddiagnostig.

The long and difficult journey many families have to go through to get a confirmed diagnosis is often called the 'diagnostic odyssey'.

Mae tri phrif reswm pam fod cyflwr genetig plentyn heb gael diagnosis:

- Mae'r cyflwr 'y prinnaf o'r prin' – cyflwr na welwyd mohono o'r blaen ac felly nid yw'n cael ei brofi.
- Mae'r cyflwr yn dangos ei hun mewn ffordd anarferol – efallai bod symptomau'r plentyn yn wahanol i blant eraill sydd â'r un cyflwr. Felly nid yw'r cyflwr yn cael ei brofi oherwydd nad yw'n ymddangos mai dyna'r cyflwr.
- Weithiau mewn profion genetig, bydd newidiadau genetig yn cael eu gweld na wyddom beth yw eu harwyddocâd clinigol, er enghraifft, newidiadau yng nghromosomau'r plentyn ond na chredir mai'r rhain sydd wedi achosi anawsterau'r plentyn. Weithiau mae hyn oherwydd bod y newid wedi'i etifeddu gan un o rieni'r plant heb fod y cyflwr wedi effeithio arno, neu oherwydd bod y deunydd genetig wedi symud o gwmpas ond yn dal yno'i gyd, mae'n ymddangos.

There are three main reasons why a child's genetic condition remains undiagnosed:

- It is the 'rarest of the rare' – a condition that hasn't been seen before and therefore isn't tested for.
- It is an unusual presentation of a known condition – the child's symptoms might be different to those of other children with the same condition. The condition is therefore not tested for because it doesn't appear to be that condition.
- Sometimes when genetic testing is undertaken genetic changes are found which are of 'unknown clinical significance', for example, changes are found in the child's chromosomes but these are not thought to be the cause of their difficulties. Sometimes this is because the change has been inherited from one of their parents who is unaffected by the condition, or because, although the genetic material has moved around, it all appears to be there.

2.8 Cael cymorth i'ch teulu ar eich taith at ddiagnosis

2.8 Accessing support for your family on your journey to diagnosis

Mae ceisio cael diagnosis i'ch plentyn yn gallu bod yn amser poenus. Mae pobl yn eich bywyd, gweithwyr iechyd proffesiynol a sefydliadau all eich helpu chi.

Trying to obtain a diagnosis for your child can be a worrying time. There are people in your life, health professionals and organisations that can help you.

Gall eich meddyg teulu, ymwelydd iechyd neu glinigydd arbenigol siarad drwy'r daith at ddiagnosis efo chi.

Gallant roi gwybodaeth i chi am y broses a gwybodaeth am ble i chwilio am gymorth.

Efallai eich bod mewn cysylltiad â Chwnselydd Genetig sy'n gallu gweithio'n uniongyrchol gyda eich teulu i gynnig gwybodaeth genetig ac a fydd yn eich cefnogi chi i wneud penderfyniadau ar gyfer eich plentyn.

Mae grwpiau cymorth i rieni hefyd a sefydliadau cymorth all roi cymorth emosiynol ac ymarferol i chi a'ch teulu.

Your GP, health visitor or specialist clinician can talk you through the journey to diagnosis.

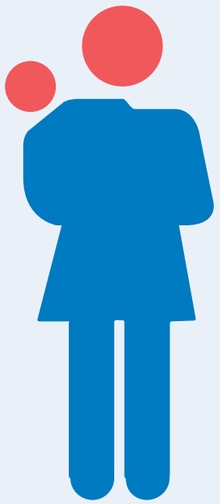
They can provide you with information on the process and information on where to seek support.

You may be in touch with a Genetic Counsellor. A Genetic Counsellor can work directly with your family to offer genetic information and will support you to make decisions for your child.

There are also parent groups and support organisations that can provide emotional and practical support for you and your family.

2.9 Dod i delerau â chael gwybod bod gan eich plentyn gyflwr genetig, prin neu heb ddiagnosis

2.9 Coming to terms with being told your child has a genetic, rare or undiagnosed condition.



Ble bynnag rydych chi ar eich taith at gael diagnosis, gall fod yn anodd dod i delerau â'r ffaith fod gan eich plentyn gyflwr iechyd.

Wherever you are on your journey to diagnosis, coming to terms with the fact that your child has a health condition can be difficult.

Mae'r newyddion bod gan eich plentyn gyflwr genetig, prin neu heb ddiagnosis yn gallu arwain at bob math o emosiynau.

'Roedd pethau'n teimlo'n llai brawychus unwaith y cawsom ddiagnosis.'
Aelod gweithdy

Mae pawb yn delio yn wahanol gyda diagnosis, neu'r diffyg diagnosis i'w plentyn. Nid oes ffordd gywir nac anghywir.

''Does gennym ddim diagnosis i fy mhlentyn. Mae'n annhebygol y cawn ni un yn fuan. Rydw i wedi dysgu canolbwyntio ar beth sydd ei angen ar fy mhlentyn rŵan a mwynhau ein bywyd gyda'n gilydd.'
Aelod gweithdy

Mae rhai rhieni yn ymateb i'r newyddion mewn ffordd debyg i brofedigaeth, mae eraill yn ymdopi drwy ddysgu pob dim y gallant am y cyflwr neu drwy ganolbwyntio eu sylw ar anghenion penodol eu plentyn.

'Wrth edrych yn ôl, mae'n debyg 'mod i wedi rhyw fath o alaru pan gawsom ddiagnosis'.
Aelod gweithdy

Weithiau, efallai byddwch chi eisiau siarad efo rhywun ynghylch sut rydych chi'n teimlo. Gallwch gysylltu â'ch meddyg teulu, eich canolfan i ofalwyr neu grŵp cymorth lleol i gael cyngor am yr help a'r cwnsela allant ei gynnig.

The news that your child has, or may have, a genetic, rare or undiagnosed condition can lead to a wide range of emotions.

'Things felt less scary once we got a diagnosis.'
Workshop participant

Every person deals with diagnosis, or the news that their child may remain undiagnosed, differently. There is no right or wrong way.

'We still don't have a diagnosis for my child, and it's unlikely we will get one soon. I've learnt to just focus on what my child needs now and enjoying our life together.'
Workshop participant

Some parents react to the news in a similar way to having a bereavement, others cope by learning all they can about the condition or focusing their attention on their child's specific needs.

'Looking back, I guess I went into a state of grief when we got a diagnosis.'
Workshop participant

Sometimes you might want to speak to someone about how you are feeling. You can contact your GP, your local carers centre or local support group for advice on the help and counselling they may be able to offer.

2.10 Beth nesaf?

2.10 What next?

Rydw i wedi cael gwybod bod gan fy mhlentyn gyflwr genetig – beth ydw i'n ei wneud nawr?

I have been told my child has a rare genetic condition - what do I do now?

Wrth gael diagnosis a gwybod enw'r cyflwr, efallai y gallwch gael gwybodaeth a chymorth i'ch helpu chi ddeall mwy am y cyflwr a sut y bydd yn effeithio ar eich plentyn.

Mae cael diagnosis o gyflwr prin iawn yn gallu bod yn rhwystredig iawn.

Efallai mai prin iawn fydd yr wybodaeth sydd ar gael am y cyflwr, ac efallai mai ychydig iawn fydd gweithwyr gofal iechyd proffesiynol a darparwyr gofal yn ei ddeall amdano. Weithiau, ni fydd ateb i lawer o gwestiynau.

Gallwch gael mwy o wybodaeth ar sut i ddelio gyda diagnosis a ble i gael gwybodaeth a chyngor yn yr Adnoddau Prin Cymorth a gwybodaeth i rieni a gofalwyr a Chymorth a gwybodaeth i'ch plentyn.

By having a diagnosis, and knowing the name of the condition, you may be able to access information and support to help you understand more about the condition and how it will affect your child.

A diagnosis of a very rare condition can be frustrating.

There may be little information on the condition available and little understanding of the condition amongst health professionals and care providers. Sometimes many questions can remain unanswered.

You can find further information on how to deal with a diagnosis and where to access information and support in the Support and information for parents and carers and the Support and information for your child Rare Resources.

Rydw i wedi cael gwybod efallai na cha' i fyth ddiagnosis i gyflwr fy mhlentyn – beth ydw i'n ei wneud rŵan?

Ni fydd nifer sylweddol o blant sy'n cael profion genetig yn cael diagnosis. Gall hyn fod yn anodd iawn ac mae'n bwysig eich bod yn cael cymorth.

SWAN UK yw'r unig rwydwaith cymorth penodol sydd ar gael i deuluoedd plant ac oedolion ifanc sydd â chyflyrau genetig heb ddiagnosis yn y DU.

Mae SWAN UK yn cefnogi cymuned o deuluoedd a phlant sydd â chyflyrau genetig heb ddiagnosis ac yn darparu ystod o wybodaeth i deuluoedd.

Cewch fwy o wybodaeth ynglŷn â lle i gael gwybodaeth a chymorth yn yr Adnoddau Prin Cymorth a gwybodaeth i rieni a gofalwyr a Chymorth a gwybodaeth i'ch plentyn.

I have been told I may never get a diagnosis for my child's condition – what do I do now?

A significant number of children having genetic testing will not receive a diagnosis. This can be very difficult and it is important you have support.

SWAN UK is the only dedicated support network available for families of children and young adults with undiagnosed genetic conditions in the UK.

SWAN UK supports a community of families of children affected by undiagnosed genetic conditions and provides a range of information for families.

You can find further information on where to access information and support in the Support and information for parents and carers and the Support and information for your child Rare Resources.

2.11 Ar ôl y diagnosis – ble i gael gwybodaeth a chymorth

2.11 After diagnosis – where to find information and support

Pan gaiff eich plentyn ddiagnosis o gyflwr genetig ac/neu brin, efallai y gwelwch nad oes llawer o wybodaeth ar gael.

When your child receives a diagnosis of a genetic and/or rare condition, you may find that there is not much information available.

Dylai eich meddyg teulu neu glinigydd arbenigol roi gwybodaeth i chi adeg y diagnosis, hyd yn oed os nad oes fawr o wybodaeth ar gael.

Gallant roi taflen wybodaeth i chi, gwybodaeth wedi'i hargraffu oddi ar y we, neu eich cyfeirio at wybodaeth briodol.

Os na ddarperir gwybodaeth, peidiwch â bod ofn gofyn i'ch meddyg teulu neu glinigydd arbenigol ble i gael gwybodaeth ddibynadwy.

Gall eich Cyngor Iechyd Cymuned helpu hefyd. Mae grwpiau i gleifion yn aml yn gallu bod yn lle gwerthfawr i gael gwybodaeth am gyflwr penodol.

Ble alla' i chwilio am grŵp i gleifion?

Gall fod yn gysur ac yn help siarad efo rhywun sydd â'r un cyflwr, neu sydd â phlentyn â'r un cyflwr – rhywun sydd â phrofiad o'r un pethau rydych chi'n eu hwynebu.

Mae byw gyda chyflwr prin (ac mae'r rhan fwyaf o gyflyrau genetig yn rhai prin) yn gallu bod yn beth unig iawn oherwydd nad oes gan lawer o bobl eraill yr un cyflwr. Mae'r un yn wir am fod yn rhiant, yn ofalwr neu'n aelod teulu i rywun sydd â chyflwr genetig, prin neu heb ddiagnosis.

Genetic Alliance UK

Gallwn helpu i gael cymorth i chi. Ar wefan Genetic Alliance UK, gallwch ddefnyddio'r botwm chwilio ar y dudalen i'r aelodau i chwilio am grwpiau all fod yn berthnasol i chi yn y DU.

Gallwch hefyd roi cynnig ar chwilio ar y we, er enghraifft, gallech geisio chwilio am enw eich cyflwr a'r geiriau 'patient support UK' mewn peiriant chwilio ar y rhyngrwyd – ond byddwch yn ofalus, mae'n hawdd i chi ddychryn eich hun oherwydd yn aml gall y canlyniadau ddangos y sefyllfa waethaf i chi.

Weithiau, ni fydd hi'n bosib dod o hyd i sefydliad sy'n benodol i gyflwr eich plentyn neu oedolyn ifanc, yn enwedig yn achos cyflyrau prin iawn.

Your GP or specialist clinician should provide you with information at the time of diagnosis, even if there is very little information available.

They may be able to provide you with a leaflet, or a printout from the internet, or they may be able to sign post you to appropriate information.

If information is not provided, don't be scared to ask your GP or specialist clinician where you can find reliable information.

Your local Community Health Council may also be able to help you. Patient groups often provide a valuable source of condition-specific information.

Where can I find a patient group?

It can be comforting and helpful to talk to someone with, or who has a child with the same condition – someone who has experienced the same things that you are facing.

Having a rare condition (which most genetic conditions are) can be extremely isolating because there aren't many other people out there living with the condition. The same goes for being a parent, carer or family member to someone with a genetic, rare, or undiagnosed condition.

Genetic Alliance UK

We can help you find support. On the Genetic Alliance UK website, you can use the 'search' function on the members' page to find groups that may be relevant to you in the UK.

You can also try an internet search, for example, it may be useful to you to search the name of your condition and the words 'patient support UK' in an internet search engine – be careful though as it is easy to scare yourself as often the results you find might be the worst case scenario.

Sometimes, particularly in the case of very rare conditions, it may not be possible to find an organisation specifically for your child or young adult's condition.

Nid yw hyn yn golygu nad oes cymorth ar gael ac efallai y byddwch chi eisiau ystyried:

Sefydliadau ymbarél

Yn aml, mae cyflyrau yn perthyn i gategorïau ehangach. Er enghraifft, mae clefyd Danon yn eithriadol o brin ac felly nid oes grŵp cymorth penodol i'r cyflwr er mwyn i deuluoedd chwilio am wybodaeth neu gyfarfod ag eraill sydd â'r cyflwr. Ond, mae'r cyflwr yn gyflwr metabolig felly gall pobl sydd â chlefyd Danon gael cymorth a gwybodaeth gan sefydliad ymbarél fel MSUK (Metabolic Support UK).

Os ydych chi eisiau gwybod i ba fath o gategori y mae cyflwr eich plentyn neu oedolyn ifanc yn perthyn, mae'n syniad da holi eich gweithwyr iechyd proffesiynol.

Fforymau ar-lein

Weithiau, nid yw teuluoedd yn gallu dod o hyd i grŵp cymorth penodol.

Nid yw hynny'n golygu na allwch chi gael cymorth. Mae'r we yn gartref i gyfoeth o lefydd rhithiol i bobl gyfarfod a siarad am eu profiadau efo clefydau prin.

Facebook

Yn llwyfan anffurfiol sy'n rhad ac am ddim, mae Facebook yn gartref i nifer o grwpiau cymorth ar-lein.

Nid oes angen i chi gael cyfrif Facebook i chwilio am grwpiau, ond bydd angen i chi gofrestru i ymuno a siarad â phobl os byddwch chi'n dod o hyd i grŵp perthnasol.

Os nad oes grŵp Facebook ar gyfer y cyflwr, gallech chi ddechrau un ar gyfer pwy bynnag fydd yn chwilio am un y tro nesaf. Gall fod yn ffordd wych i gysylltu ag eraill yn uniongyrchol.

Cymuned Ar-lein Contact

Mae Cymuned Ar-lein Contact yn fforwm ar-lein sydd wedi'i ffurfio gan Contact i rieni plant sydd ag anableddau.

Teipiwch eich cyflwr yn y bocs chwilio ar y wefan i weld faint o bobl sydd wedi cofrestru dan enw'r cyflwr.

Cofiwch gofrestru eich hun, hyd yn oed os nad oes neb arall wedi cofrestru, fel bod unrhyw un sydd newydd gael diagnosis yn gallu dod o hyd i chi. bit.ly/rrcontact

This does not mean there is no support available and you may wish to consider:

Umbrella organisations

Often conditions fall into wider categories of conditions. For example, Danon disease is extremely rare and therefore does not have a condition specific support group for families to find information or meet others with Danon disease. But, Danon disease is a metabolic condition, so people with Danon disease can access support and information from an umbrella organisation such as MSUK (Metabolic Support UK).

If you want to know what type of category your child or young adult's condition falls under, it is a good idea to ask your health professional.

Online Forums

Sometimes families are unable to find a specific support group.

That doesn't mean that you can't find support. The internet is home to a wealth of virtual areas for people to meet and talk about their experiences with rare disease.

Facebook

As a free, informal platform Facebook is home to a number of online support groups.

You don't need to have a Facebook account to search for groups, although if you do find a relevant group you will need to sign up to join and talk to people.

If there isn't already a Facebook group for the condition you can always set one up for the next person searching to find. It can be a great way to connect with others directly.

Contact Online Community

Contact Online Community is an online forum set up by the organisation Contact for parents of children with disabilities.

Just enter your condition into the search function on the website and you'll be able to see how many people have registered under the disease name.

Don't forget to register yourself, even if there isn't anyone else registered, so that anyone who is newly diagnosed can find you. bit.ly/rrcontact

Mwy o wybodaeth

SWAN UK: bit.ly/rrjoinswanuk
Contact: bit.ly/rrpatternsofinheritance
Cymorth a gwybodaeth SWAN UK:
bit.ly/rrswanuksupportandinformation
Unique: bit.ly/rrunique

Mae'r grŵp cymorth Unique yn cynnig gwybodaeth a chymorth i deuluoedd sydd ag anhwylderau cromosom prin a, ble'n bosibl, yn cysylltu teuluoedd plant sydd â'r un cyflwr prin. bit.ly/rrunique

Awgrymiadau

Cewch fanylion am y ganolfan i ofalwyr neu'r grwpiau cymorth sy'n lleol i chi yn y 'Cyfeiriadur Gwybodaeth' o fewn yr Adnoddau Prin.

Further information

SWAN UK: bit.ly/rrjoinswanuk
Contact: bit.ly/rrpatternsofinheritance
SWAN UK support and information:
bit.ly/rrswanuksupportandinformation
Unique: bit.ly/rrunique

The support group Unique offers information and support to families with rare chromosome disorders and, where possible, will link together families whose children have the same rare condition. bit.ly/rrunique

Hints

You can find details of local carer's centre or local support groups in the Rare Resources Information Directory.

Cyngor Gan Rieni - delio gyda diagnosis

Cymryd eich amser – Nid oes angen i chi ofyn pob cwestiwn na dysgu popeth am y cyflwr i gyd ar unwaith. Cymerwch eich amser i brosesu'r wybodaeth yn eich amser eich hun a meddwl am eich teimladau.

Siarad – Siaradwch â rhywun sy'n agos atoch chi am y diagnosis ac am sut rydych chi'n teimlo. Os nad ydych chi'n teimlo y gallwch siarad â rhywun rydych chi'n 'nabod, yna rhowch gynnig ar sefydliad sydd â llinell gymorth (cewch fanylion yn y Cyfeiriadur Gwybodaeth).

Chwilio am grŵp cymorth – Efallai bod grŵp cymorth ar gael i rai cyflyrau. Gall grwpiau cymorth roi gwybodaeth am gyflwr a chymorth i'ch teulu.

Ymchwilio – Mae rhai pobl yn gweld ei bod hi'n fuddiol chwilio am wybodaeth am y cyflwr ac ymchwilio i'r cyflwr.

Siarad â theuluoedd eraill – Mae rhai pobl yn gweld ei bod hi'n gysur ac yn fuddiol siarad â theuluoedd eraill sydd â phlentyn â chyflwr tebyg.

Bod yn bositif – Cofiwch ganolbwyntio ar yr hyn sy'n dda am fywyd efo eich plentyn. Mae diagnosis yn gallu llethu rhywun, felly treuliwch amser yn canolbwyntio ar y pethau da.

Top Tips From Parents - dealing with diagnosis

Take your time – You don't need to ask every question or learn everything about the condition all at once. Take your time to process information at your own pace and think about your feelings.

Talk – Talk to someone you are close to about the diagnosis and how you are feeling. If you don't feel that you can speak to someone you know, try an organisation that provides a support helpline (find details in the Information Directory).

Find a support group – For some conditions there may be a support group available. Support groups can provide information on a condition and support for your family.

Research – For some people, it can be helpful to find information on and research the condition.

Speak to other families – For some people, speaking to other families who have a child with a similar condition can be reassuring and helpful.

Be positive – Remember to focus on the good things about life with your child. It can be easy to become overwhelmed by a diagnosis, so spend time focusing on positives.

Cyngor ar gael gwybodaeth ddibynadwy

Edrych am y logo safonau gwybodaeth – Mae'r safonau gwybodaeth yn rhaglen er mwyn rheoleiddio gwybodaeth am iechyd a gofal. Os yw'r sefydliad yn dangos y logo safonau gwybodaeth, yna mae'r wybodaeth honno'n ddibynadwy.

Arbenigwr ar y cyflwr – Wrth chwilio am wybodaeth am gyflwr prin iawn, gall fod yn help chwilio am arbenigwr ar y cyflwr. Bydd arbenigwyr yn aml yn ysgrifennu erthyglau ar gyflyrau mewn cyfnodolion, yn rhoi manylion achosion y maent wedi'u gweld, ffyrdd y maent wedi trin symptomau ac ati. Efallai y byddwch chi eisiau chwilio am awduron papurau ar y cyflwr, gan mai nhw fydd yr arbenigwyr ar y cyflwr.

Awgrym gwych ar gyfer chwilio am wybodaeth

Gofalwch eich bod chi'n gwybod yr holl enwau y mae pobl yn eu defnyddio am gyflwr – gallwch wneud hyn drwy fynd ar Orphanet a chwilio am eich cyflwr. Pan fyddwch chi wedi dod o hyd i'r dudalen iawn, dylech weld bod rhestr o enwau eraill ar y cyflwr yn y bocs glas.

Mwy o wybodaeth

Mae Orphanet yn wefan Ewropeaidd sy'n rhoi gwybodaeth am gyflyrau prin. bit.ly/rrorphanet

Tips for finding reliable information

Look for the information standards logo – The information standard is a programme set up to regulate health and care information. If an organisation is displaying the information standards logo you know you can trust the information.

Specialist in the condition - When trying to find information on a really rare condition it may be helpful to find a specialist in the condition. Experts often write articles for journals about conditions, detailing cases they have seen, ways they have treated symptoms etc. You may want to look for authors of papers on the condition, as they will be the expert on the condition.

Top tip for finding Information

Make sure you know all the names that people use for a condition - you can do this by going to orphanet and searching for your condition. When you have found the right page you should see that in the blue table there is a list of other names for the condition.

Further information

Orphanet is a European website providing information about rare diseases. bit.ly/rrorphanet

Eich **Nodiadau**

Your **Notes**


GENETIC
ALLIANCE UK