RARE resources WALES

7 Canllaw i Deuluoedd

7 Families' Guide

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

Er mwyn gofyn am gopi caled o unrhyw un o'r canllawiau Rare Resources, cysylltwch â Genetic Alliance UK ar contactus@geneticalliance.org.uk neu 0300 124 0441.

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

To request a hard copy of any of the Rare Resources guides, please contact Genetic Alliance UK at contactus@geneticalliance.org.uk or 0300 124 0441.

GENETIC ALLIANCE UK

RARE resources WALES

Canllaw i rieni sydd â phlentyn neu berson ifanc yn byw efo cyflwr genetig, prin neu heb ddiagnosis yng Nghymru.

A guide for families with a child or young person living with a genetic, rare or undiagnosed condition in Wales.

7.1 I bwy mae'r canllaw hwn?

7.1 Who is this guide for?

Os ydych chi wedi codi'r daflen hon neu wedi'i chael gan rywun sy'n rhoi gofal i'ch plentyn, mae'n debyg eich bod chi'n rhiant/gofalwr i blentyn neu berson ifanc sydd â/allai fod â chyflwr genetig, prin neu heb ddiagnosis.

If you have picked up this leaflet, or have been given it by someone involved in your child's care, it is likely that you are the parent carer of a child or young person who has, or may have, a genetic, rare or undiagnosed condition.

Mae'r canllaw yn rhoi cyflwyniad byr i'r hyn y mae byw gyda cyflwr genetig, prin neu heb ddiagnosis yn ei olygu, ac mae'n rhoi dolenni defnyddiol at wybodaeth a chymorth sy'n ddibynadwy. Cynhyrchwyd y canllaw hwn gan Genetic Alliance UK, ar y cyd â theuluoedd yng Nghymru.

Genetic Alliance UK yw'r elusen genedlaethol sydd yn gweithio i wella bywydau cleifion a theuluoedd y mae cyflyrau prin, genetig a heb ddiagnosis yn effeithio arnynt. Mae mwy na 200 o gyrff cleifion yn perthyn i'n cynghrair ni.

Rydym yn gartref i Rare Disease UK – yr ymgyrch genedlaethol i bobl sydd â chyflyrau prin ac sy'n eu cefnogi – a SWAN UK (syndromes without a name), yr unig rwydwaith cymorth penodol sydd ar gael i deuluoedd plant ac oedolion ifanc sydd â chyflyrau genetig heb ddiagnosis yn y DU.

Beth yw cyflyrau genetig, prin a heb ddiagnosis?

Mae cyflyrau genetig, prin a heb ddiagnosis yn cynnwys amrywiaeth fawr o gyflyrau iechyd a byddant hefyd yn amrywio o ran eu difrifoldeb a'r effaith a gânt.

Mae llawer o wahanol fathau o gyflyrau genetig a phrin, a bydd y ffordd y bydd y cyflwr yn effeithio ar deulu yn dibynnu ar nifer o ffactorau. Er hynny, nid yw'n anghyffredin i deuluoedd plant a phobl ifanc sydd â chyflyrau genetig, prin a heb ddiagnosis wynebu'r un math o heriau.

Mae'r rhain yn aml yn cynnwys:
- Taith hir at ddiagnosis
- Ansicrwydd am y dyfodol
- Diffyg dealltwriaeth gan weithwyr proffesiynol sy'n rhan o ofal eu plentyn.
- Gofal wedi'i drefnu'n wael, llawer o wahanol

This guide provides a brief introduction to what it means to have a genetic, rare or undiagnosed condition and provides useful links to where you can find sources of reliable information and support. This guide has been produced by Genetic Alliance UK, in collaboration with families in Wales.

Genetic Alliance UK is the national charity working to improve the lives of patients and families affected by rare, genetic and undiagnosed conditions. We are an alliance of over 200 patient organisations.

We are home to Rare Disease UK – the national campaign for people with rare diseases and all who support them – and SWAN UK (syndromes without a name), the only dedicated support network available for families of children and young adults with undiagnosed genetic conditions in the UK.

What are genetic, rare and undiagnosed conditions?

Genetic, rare and undiagnosed conditions cover a broad range of health conditions that will also vary in their severity and the impact they have.

There are many types of genetic and rare conditions and how a condition affects a family will depend on a number of factors, however it is not uncommon for families of children and young people with rare, genetic and undiagnosed conditions to experience similar challenges.

These often include:
- A long journey to diagnosis.
- Uncertainty about the future.
- Lack of understanding from professionals involved in the care of their child.
- Poorly coordinated care with lots of different health care professionals involved and lots of appointments.

weithwyr gofal iechyd proffesiynol yn rhoi gofal a llawer o apwyntiadau.
- Trafferth cael gwybodaeth a chymorth gan arbenigwr, neu grŵp i gleifion, ar gyfer cyflwr eu plentyn.
- Teimlo'n unig.

Mae cyflwr genetig yn cael ei achosi gan newidiadau yn nilyniant DNA unigolyn. Mae gan bawb ohonom lawer o newidiadau yn ein dilyniant DNA ac nid yw'r rhain yn cael unrhyw effaith yn aml. Ond weithiau, gall y newidiadau hyn arwain at anableddau dysgu, oediad datblygiadol neu broblemau iechyd eraill. Gall cyflyrau genetig fod yn anhwylderau un gennyn, cromosomol neu gymhleth.

Cyflwr prin yw unrhyw gyflwr sy'n effeithio ar lai na 5 mewn 10,000 o bobl. Mae rhwng 6,000 ac 8,000 o gyflyrau prin. Weithiau, efallai bod amheuaeth fod gan blentyn neu berson ifanc gyflwr genetig, er bod y profion genetig, hyd yma, wedi methu adnabod y newid yn y DNA sydd wedi achosi'r cyflwr. Weithiau mae cyflyrau genetig heb ddiagnosis yn cael eu galw yn 'syndromau heb enw', neu '**syndromes without a name, SWAN**' yn fyr. I blant sydd â syndrom heb enw, maent yn gallu cael gwahanol fathau o symptomau ac mae'r syndrom yn debygol o effeithio'n wahanol ar bob plentyn. Gallwch ddarllen mwy am gyflyrau genetig, prin a heb ddiagnosis yma:

Genetic Alliance UK - geneticalliance.org.uk
Rare Disease UK – rarediseasе.org.uk
SWAN UK - undiagnosed.org.uk

- Difficulties in accessing information and support from an expert, or patient group, for their child's condition.
- Experiencing feelings of isolation.

A genetic condition is caused by changes in an individual's DNA sequence. We all have lots of changes in our DNA sequence and often these don't have an impact. Sometimes however, these changes can result in learning disabilities, developmental delay and other health problems. Genetic conditions may be single gene, chromosomal or complex disorders.

A rare condition is any condition that affects less than 5 in 10,000 people. There are between 6,000 and 8,000 rare conditions. Sometimes a child or young person may be suspected to have a genetic condition, but genetic testing has so far failed to identify the change in the DNA that has caused it.

Sometimes undiagnosed conditions are called '**syndromes without a name**' or '**SWAN**' for short. Children affected by a syndrome without a name can have a range of different symptoms and each child is likely to be affected differently.

Find out more about genetic, rare and undiagnosed conditions:
Genetic Alliance UK – geneticalliance.org.uk
Rare Disease UK – raredisease.org.uk
SWAN UK – undiagnosed.org.uk

7.2 Ble i gael gafael ar wybodaeth sy'n ddibynadwy?

7.2 Where can I find reliable information?

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

Gall y cyrff hyn fod o gymorth i'ch helpu chi gael gwybodaeth safonol a dibynadwy:

Mae **Genetic Alliance UK** yn gynghrair genedlaethol o gyrff efo mwy na 200 o elusennau yn perthyn iddi sy'n cefnogi plant, teuluoedd ac unigolion y mae cyflyrau genetig yn effeithio arnynt. Gallwch chwilio drwy restr o'r cyrff sy'n aelodau ar wefan Genetic Alliance UK. Geneticalliance.org.uk

Contact yw'r elusen i deuluoedd plant anabl. Ar eu gwefan, gallwch chwilio drwy eu cyfeiriadur o gyflyrau meddygol ble cewch ddolenni at wybodaeth a chymorth. contact.org.uk/cymru/

Mae **Unique** yn rhoi cymorth i deuluoedd y mae anhwylderau cromosom prin neu anhwylderau genynnau trech awtosomaidd. Mae ganddynt ganllawiau rhad ac am ddim ar anhwylderau cromosomau a genynnau penodol. rarechromo.org

Mae **Orphanet** yn borth ar lein ble cewch wybodaeth ar gyflyrau unigol. Orpha.net

Your GP or specialist clinician should provide you with information at the time of diagnosis, even if there is very little information available. If information is not provided, don't be afraid to ask where you can access reliable information.

You may find these organisations helpful in assisting you to find good quality, reliable information:

Genetic Alliance UK is a national alliance of organisations with a membership of over 200 charities that support children, families and individuals affected by genetic conditions. You can search a list of member organisations on the Genetic Alliance UK website. Geneticalliance.org.uk

Contact is a charity for families with disabled children. Their website provides a searchable A to Z directory of medical conditions with links to information and support organisations. contact.org.uk/cymru/

Unique provides support for families affected by rare chromosome disorders or autosomal dominant gene disorders. They provide free guides on specific chromosome and gene disorders. rarechromo.org

Orphanet is an online portal that provides information on individual conditions. Orpha.net

Ble i gael cymorth?

Gall fod yn gysur ac yn fuddiol siarad â rhywun sydd â'r un cyflwr, neu sydd â phlentyn sydd â'r un cyflwr – rhywun sydd wedi cael profiad o'r un pethau rydych chi'n eu hwynebu.

Mae byw gyda chyflwr genetig, 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 berthynas i rywun sydd â chyflwr genetig, prin neu heb ddiagnosis.

Gall Genetic Alliance UK eich helpu chi i gael cymorth. 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. Weithiau, ni fydd hi'n bosib dod o hyd i sefydliad sy'n benodol i gyflwr eich plentyn, yn enwedig yn achos cyflyrau prin iawn. 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. Ond, mae'r cyflwr yn gyflwr metabolig felly gall pobl sydd â chlefyd Danon gael cymorth a gwybodaeth gan sefydliad ymbarél fel Climb (Children living with Inherited Metabolic Conditions). 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.

Where can I find support?

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 you are facing.

Having a rare, genetic condition can be isolating because there aren't many other people 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 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. Sometimes, particularly in the case of very rare conditions, it may not be possible to find an organisation specifically for your child's condition. This does not mean that there is not support and you may wish to consider umbrella organisations.

Often conditions fall within wider categories of conditions. For example, Danon disease is extremely rare and doesn't have a specific support group. But Danon disease is a metabolic condition, so people can access support and information from the umbrella organisation Climb (Children living with Inherited Metabolic Conditions). 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.

7.3 **Y Pecyn Adnoddau Prin**

7.3 **The Rare Resources Toolkit**

Canllaw yw Adnoddau Prin sydd wedi'i gynhyrchu gan Genetic Alliance UK i deuluoedd sydd wedi cael diagnosis o gyflwr genetig neu brin yn ddiweddar, sydd ar y daith at gael diagnosis, neu sydd wedi cael gwybod bod cyflwr eu plentyn mor brin na chaiff ddiagnosis, o bosib.

Rare Resources is a guide produced by Genetic Alliance UK for families who have recently received a diagnosis of a genetic or rare condition, are on the journey to a diagnosis, or who have beentold their child's condition is so rare they might not get a diagnosis.

Yn y Pecyn, mae amrywiaeth fawr o wybodaeth gyffredinol am gyflyrau prin, genetig a heb ddiagnosis yn ogystal â sut i gael gafael ar wybodaeth, gofal a chymorth sy'n ddibynadwy. I ofyn am gopi papur o'r Pecyn Adnoddau Prin, cysylltwch â Genetic Alliance UK contactus@geneticalliance.org.uk

Yn y Pecyn, mae'r adrannau hyn:
- Esbonio cyflyrau genetig, prin a heb ddiagnosis
- Y daith at ddiagnosis – Cymorth a gwybodaeth i rieni a gofalwyr
- Cymorth a gwybodaeth i'ch plentyn
- Defnyddio'r GIG yng Nghymru
- Cyfeiriadur gwybodaeth yn manylu ar y gwasanaethau cymorth sydd ar gael yng Nghymru

Gallwch lawr lwytho'r Pecyn o bit.ly/raresourceswales

The Toolkit contains a wide range of general information on rare, genetic and undiagnosed conditions as well as information on how to access reliable information, care and support. To request a hard copy of the Rare Resources Toolkit, please contact Genetic Alliance UK contactus@geneticalliance.org.uk

The Toolkit contains the following sections:
- Genetic, rare and undiagnosed conditions explained
- The journey to diagnosis – Support and information for parents and carers
- Support and information for your child
- Using the NHS in Wales
- Information directory detailing support services available in Wales

The Toolkit can be downloaded from bit.ly/raresourceswales

Cyngor

gan deuluoedd, i deuluoedd

Datblygwyd y Pecyn Adnoddau Prin ochr yn ochr â rhieni sy'n ofalwyr a gweithwyr iechyd proffesiynol sydd yn gofalu am blant sydd â chyflwr genetig, prin neu heb ddiagnosis. Dyma rai pytiau o 'gyngor' ganddynt:

Chwilio am wybodaeth a chymorth sy'n ddibynadwy

Gall fod yn fuddiol chwilio am wybodaeth am gyflwr eich plentyn. Efallai bod grŵp cymorth ar gael ar gyfer rhai cyflyrau. Mae'r Pecyn Adnoddau Prin yn rhoi gwybodaeth ar sut i fynd at wybodaeth a chymorth sy'n ddibynadwy, hyd yn oed os nad yw eich plentyn wedi cael diagnosis. Peidiwch â bod ofn gofyn am help – cofiwch, mae'n berffaith naturiol i chi deimlo bod pethau'n ormod i chi ar brydiau. Peidiwch â bod ofn gofyn am help os oes ei angen arnoch.

Cyfarfod â rhieni eraill sy'n ofalwyr

Gallwch elwa llawer drwy gyfarfod ag eraill sydd yn, neu wedi, cael yr un profiad â chi. Mae'r Pecyn Adnoddau Prin yn egluro sut allwch chi chwilio am gymorth gan deuluoedd eraill

Deall eich hawliau, a hawliau eich plentyn

Efallai y gwelwch chi fod gennych chi a'ch plentyn yr hawl i gael cymorth ymarferol, emosiynol ac ariannol. Mae'r Pecyn yn egluro'r cymorth sydd ar gael i deuluoedd yng Nghymru.

Rhoi trefn ar bethau

Cadwch gopi o'r holl lythyrau a'r wybodaeth sy'n ymwneud ag iechyd eich plentyn efo'i gilydd. Fel hyn, bydd yr wybodaeth bwysig i gyd mewn un lle ac yn eich helpu chi i gadw golwg ar gynnydd eich plentyn. Mae'r Pecyn yn rhoi cyngor ar reoli apwyntiadau eich plentyn a defnyddio'r GIG yng Nghymru.

Top Tips

from families, for families

The Rare Resources Toolkit has been developed in collaboration with parent carers and health professionals involved in the care of children with a genetic, rare or undiagnosed condition. Here are some 'top tips' from them.

Find reliable information and support

It may be helpful to find information on your child's condition. For some conditions there may be a support group available. The Rare Resources Toolkit provides information on how to access reliable information and support, even if your child does not have a diagnosis. Don't be afraid to ask for help – Remember it is completely normal to feel overwhelmed from time to time. Don't be afraid to ask for help if you need it.

Meet other parent carers

It can be very beneficial to meet others who are experiencing, or have experienced what you are going through. The Rare Resources Toolkit explains how you can find support from other families.

Understand your rights, and the rights of your child

You may find that you and your child are eligible for practical, emotional and financial support. The Rare Resources Toolkit explains support available to families in Wales.

Get organised

Keep a copy of all letters and information relating to your child's health together. This will ensure you have the important information together and help you keep track of your child's progress. The Rare Resources Toolkit provides advice on managing your child's appointments and using the NHS in Wales.

Eich **Nodiadau**

Your **Notes**


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