RARE resources WALES

1 Eglurhad o gyflyrau genetig, prin a heb ddiagnosis

1 Genetic, rare and undiagnosed conditions explained

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

1.1 Beth yw cyflyrau genetig, prin a heb ddiagnosis?

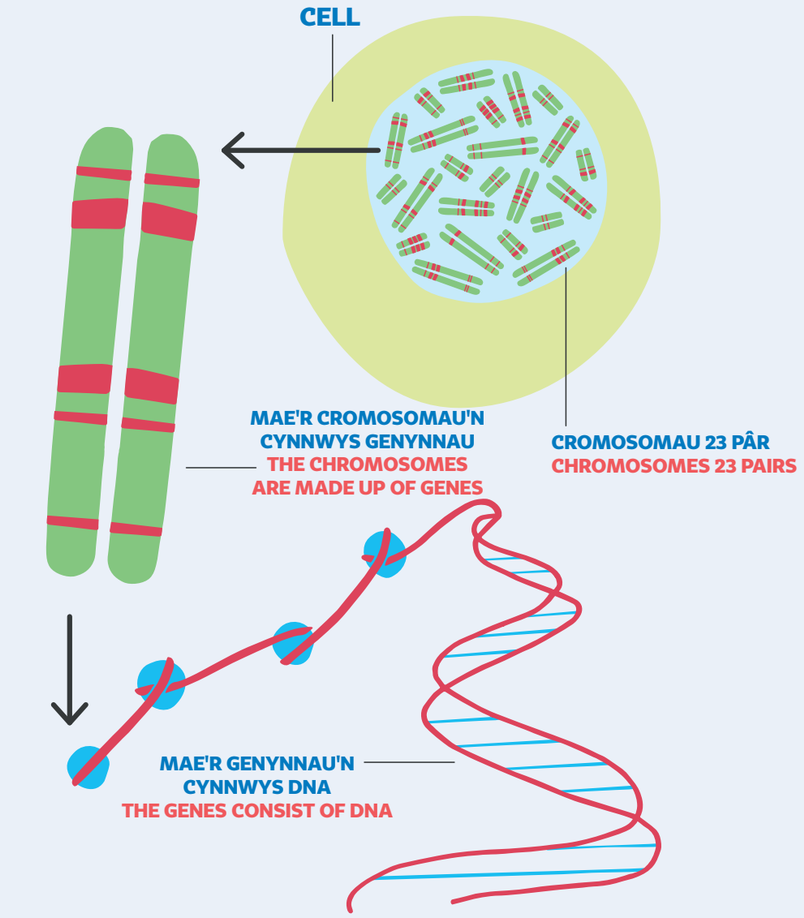
1.2 Beth mae'n ei olygu i gael cyflwr genetig, prin neu heb ddiagnosis?

1.1 What are genetic, rare or undiagnosed conditions?

1.2 What does it mean to have a genetic, rare or undiagnosed condition?

1.1 Beth yw cyflyrau genetig, prin neu heb ddiagnosis?

1.1 What are genetic, rare or undiagnosed conditions?



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. Er mwyn deall beth yw cyflyrau genetig, prin a heb ddiagnosis, mae'n rhaid i ni yn gyntaf oll ddeall rôl genynnau a chromosomau.

Genetic, rare and undiagnosed conditions cover a broad range of health conditions that will also vary in their severity and the impact they have. To understand what genetic, rare and undiagnosed conditions are, first we must understand the role of genes and chromosomes.

Genynnau a chromosomau

Mae miliynau o gelloedd yn ein corff. Mae'r rhan fwyaf o gelloedd yn cynnwys set gyflawn o enynnau. Mae genynnau fel set o gyfarwyddiadau, sy'n rheoli ein tyfiant a sut mae ein cyrff yn gweithio. Nhw hefyd sy'n gyfrifol am lawer o'n nodweddion, fel lliw ein llygaid, ein teip gwaed a'n taldra. Mae gennym filoedd o enynnau.

Mae genynnau i'w cael ar strwythurau bach tebyg i edafedd o'r enw cromosomau. Fel arfer, mae gennym 46 o gromosomau yn y rhan fwyaf o gelloedd. Rydym yn etifeddu un set o 23 cromosom gan ein mam ac un set o 23 cromosom gan ein tad. Felly, mae gennym ddwy set o 23 cromosom, neu 23 pâr.

Oherwydd bod y cromosomau wedi'u gwneud o enynnau, rydym yn etifeddu dau gopi o'r rhan fwyaf o enynnau, un copi gan bob rhiant. Dyma'r rheswm fod gennym nodweddion tebyg i'n rhieni. Mae'r cromosomau, ac felly'r genynnau, wedi'u gwneud o sylwedd cemegol o'r enw DNA.

Mae cromosomau rhif un i 22 yn edrych yr un fath mewn merched a dynion. Awtosomau yw'r enw ar y rhain.

Mae pâr rhif 23 yn wahanol mewn dynion a merched a'r cromosomau rhyw yw'r enw ar y rhain. Mae dau fath o gromosomau rhyw, sef cromosom-X a chromosom-Y.

Fel arfer, mae gan ferched ddau gromosom X (XX). Mae merch yn etifeddu un cromosom X gan ei mam ac un cromosom X gan ei thad. Mae gan ddynion fel arfer cromosom X a chromosom Y (XY). Mae bachgen yn etifeddu cromosom X gan ei fam a chromosom Y gan ei dad.

Mae'n bwysig bod gennym ddigon o ddefnydd cromosom, gan fod y genynnau (sy'n rhoi cyfarwyddiadau i'r celloedd yn ein corff) i'w cael ar y cromosomau.

Genes and chromosomes

Our bodies are made up of millions of cells. Most cells contain a complete set of genes. Genes act like a set of instructions, controlling our growth and how our bodies work.

They are also responsible for many of our characteristics, such as our eye colour, blood type and height. We have thousands of genes.

Genes are located on small thread-like structures called chromosomes. Usually we have 46 chromosomes in most cells. We inherit one set of 23 chromosomes from our mother and one set of 23 chromosomes from our father. So we have two sets of 23 chromosomes, or 23 pairs.

Because the chromosomes are made up of genes, we inherit two copies of most genes, one copy from each parent. This is the reason we often have similar characteristics to our parents. The chromosomes, and therefore the genes, are made up of a chemical substance called DNA.

The chromosomes numbered one to 22 look the same in males and females. These are called the autosomes.

Pair number 23 is different in males and females and they are called the sex chromosomes. There are two kinds of sex chromosomes, the X chromosome and the Y chromosome.

Females normally have two X chromosomes (XX). A female inherits one X chromosome from her mother and one X chromosome from her father. Males normally have an X and a Y chromosome (XY). A male inherits an X chromosome from his mother and a Y chromosome from his father.

It is important that we have the correct amount of chromosome material, as the genes (that instruct the cells in our body) are found on the chromosomes.

Cyflyrau genetig

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. Mae modd rhannu cyflyrau genetig yn dri chategori gwahanol: anhwylderau un gennyn, cromosomol neu gymhleth

Mae anhwylderau un gennyn yn cael eu hachosi gan newidiadau mewn un gennyn neilltuol.

Mae mwy na 10,000 o anhwylderau dynol sy'n cael eu hachosi gan newid mewn un gennyn. Ar eu pen eu hunain, mae pob anhwylder un gennyn yn brin iawn, ond ar y cyfan, maent yn effeithio ar ryw un y cant o'r boblogaeth.

Gan mai dim ond un gennyn sydd dan sylw, mae'n hawdd olrhain yr anhwylderau hyn drwy deuluoedd yn aml. Gall genetegwyr ragdybio'r risg iddynt ddigwydd mewn cenedlaethau yn y dyfodol. Mae modd rhannu anhwylderau un gennyn yn wahanol gategorïau: dominyddol (dominant), enciliol (recessive) a chysylltiedig ag x (x-linked).

Mae enghreifftiau o anhwylderau un gennyn yn cynnwys ffeibrosis systig, clefyd y crymangelloedd, syndrom X frau, nychdod cyhyrol a chlefyd Huntington.

Mae anhwylderau cromosomol yn digwydd o ganlyniad i newidiadau yn nifer neu strwythur y cromosomau.

Mae newidiadau yn nifer y cromosomau yn digwydd pan fydd mwy na'r arfer o gopïau o gromosom neilltuol, neu lai ohonynt. Mae newidiadau yn strwythur y cromosom yn digwydd pan fydd y deunydd mewn un cromosom wedi'i ddrysu neu ei aildrefnu mewn rhyw ffordd. Gall hyn olygu bod rhannau o gromosom wedi'u hychwanegu neu eu colli.

Enghreifftiau o anhwylderau cromosomol yw syndrom Down neu drisomedd 21, syndrom Cri du chat, syndrom Turner a syndrom Wolf-Hirschhorn.

Anhwylderau cymhleth (sydd hefyd yn cael eu galw'n aml-ffactorol neu'n bolygenaidd) yw'r rheiny sy'n cael eu hachosi gan effaith llawer o wahanol enynnau ar yr un pryd, sydd yn aml yn rhyngweithio mewn ffordd gymhleth â ffactorau amgylcheddol a ffordd o fyw fel diet.

Mae elfen genetig gref i lawer o'r afiechydon cyffredin a gaiff oedolion, fel diabetes mellitus, gordyndra, sgitsoffrenia a'r annormaleddau datblygiadol mwyaf cyffredin, fel gwefus hollt a namau cynhenid ar y galon. Mae mwy nag un newid genetig yn eu hachosi.

Oherwydd bod afiechydon polygenaidd yn cynnwys mwy nag un gennyn, mae patrymau etifeddu yn amrywiol ac yn gymhleth. Os oes gan riant afiechyd, nid yw o angenrheidrwydd yn golygu y bydd plentyn yn datblygu'r un afiechyd. Ar y llaw arall, efallai na fydd unigolyn yn cael ei eni efo afiechyd ond gall y risg o'i ddatblygu fod yn uwch. Mae hyn yn cael ei alw'n dueddiad neu'n rhagdueddiad genetig.

Genetic conditions

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 any impact.

Sometimes however these changes can result in learning disabilities, developmental delay or other health problems. Genetic conditions can be divided into three different categories: single gene, chromosomal or complex disorders.

Single gene disorders are caused by changes in one particular gene.

There are over 10,000 human disorders caused by a change in a single gene. Individually, single gene disorders are each very rare, but as a whole, they affect about one percent of the population.

Since only a single gene is involved, these disorders can often be easily tracked through families, and geneticists can predict the risk of them occurring in later generations. Single gene disorders can be divided into different categories: dominant, recessive and X-linked.

Examples of single gene disorders include cystic fibrosis, sickle cell disease, fragile X syndrome, muscular dystrophy and Huntington's disease.

Chromosomal disorders result from changes in the number or structure of the chromosomes.

Changes in the number of chromosomes happen when there are more or fewer copies of a particular chromosome than usual. Changes in chromosome structure happens when the material in an individual chromosome is disrupted or rearranged in some way. This may involve the addition or loss of parts of a chromosome.

Examples of chromosomal disorders include Down's syndrome or trisomy 21, Cri du chat syndrome, Turner syndrome and Wolf-Hirschhorn syndrome.

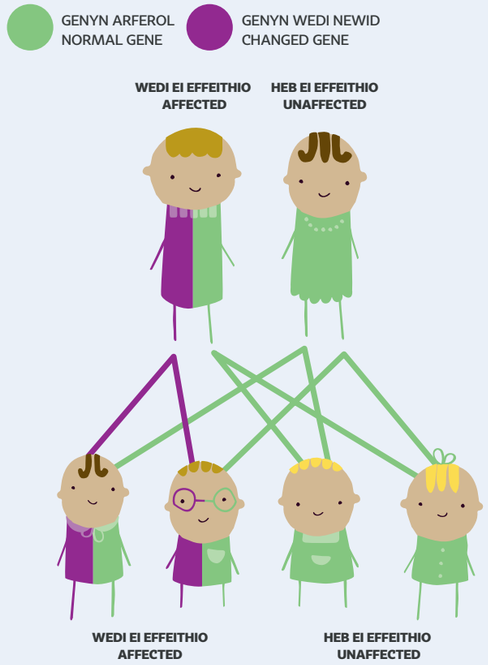
Complex disorders (also known as multifactorial or polygenic) are those that are caused by the simultaneous effect of many different genes, often in complex interaction with environmental and lifestyle factors such as diet.

Many of the common diseases of adult life, such as diabetes mellitus, hypertension, and most common developmental abnormalities, such as cleft lip and congenital heart defects, have a strong genetic component and are caused by more than one genetic change.

Because polygenic diseases involve more than one gene, inheritance patterns are diverse and complex. If a parent has a disease, it does not necessarily mean a child will develop the same disease. On the other hand, an individual may not be born with a disease but may be at a higher risk of developing it. This is known as genetic predisposition or susceptibility.

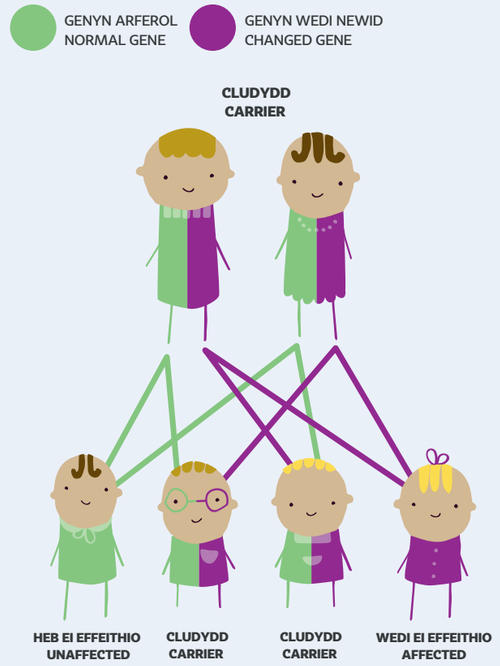
SUT MAE CYFLYRAU TRECHAF YN CAEL EU HETIFEDDU?
HOW ARE DOMINANT CONDITIONS INHERITED?

Sut mae cyflyrau trech yn cael eu trosglwyddo o riant i blentyn
How dominant conditions are passed on from parent to child

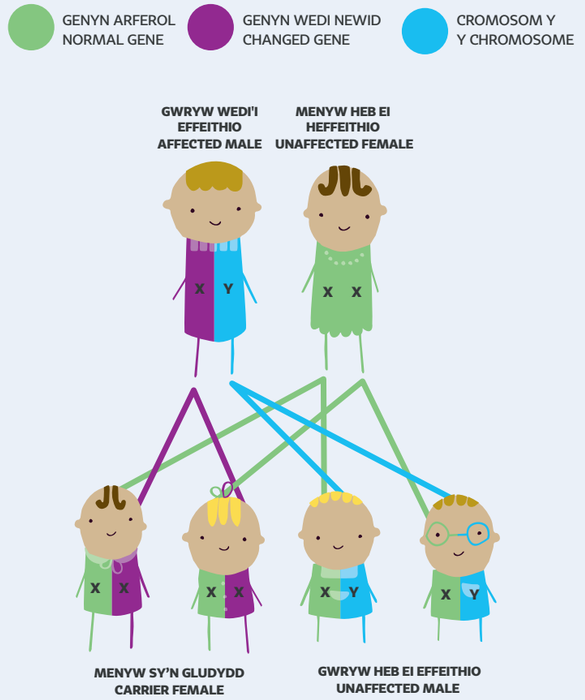


SUT MAE CYFLYRAU ENCILIOL YN CAEL EU HETIFEDDU?
HOW ARE RECESSIVE CONDITIONS INHERITED?

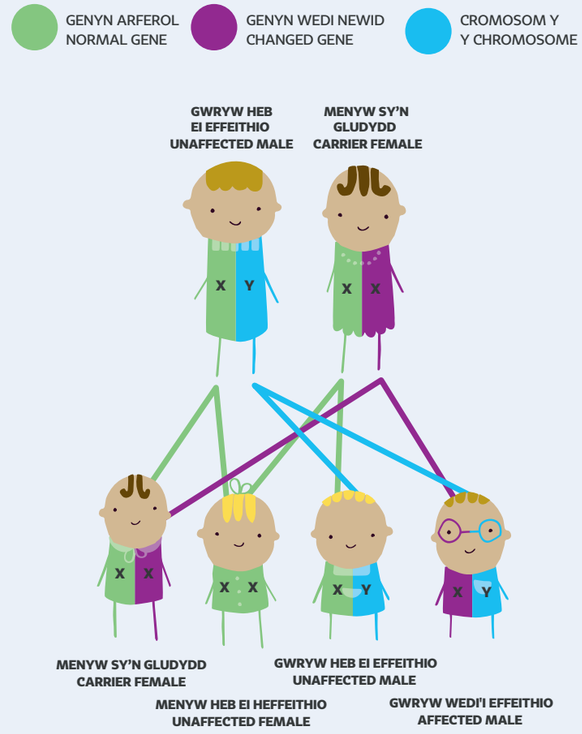
Sut mae cyflyrau enciliol yn cael eu trosglwyddo o riant i blentyn
How recessive conditions are passed on from parent to child



SUT MAE CYFLYRAU ENCILIOL SY'N GYSYLLTIEDIG AG X YN CAEL EU TROSGLWYDDO GAN DDYNION Y MAENT YN EFFEITHIO ARNYNT
HOW X-LINKED RECESSIVE CONDITIONS ARE PASSED ON BY AFFECTED MALES



SUT MAE CYFLYRAU ENCILIOL SY'N GYSYLLTIEDIG AG X YN CAEL EU TROSGLWYDDO GAN GLUDWYR BENYWAIDD
HOW X-LINKED RECESSIVE CONDITIONS ARE PASSED ON BY FEMALE CARRIERS



Cyflyrau prin

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.

Bydd cyflwr prin yn effeithio ar 1 mewn 17 o bobl, neu saith y cant o'r boblogaeth, ar ryw adeg yn eu bywyd. Mae hyn gyfystyr â ryw 220,000 o bobl yng Nghymru. Mae'r rhan fwyaf o gyflyrau prin hefyd yn gyflyrau genetig (ac felly yn ganlyniad i newid yn nilyniant DNA unigolyn).

Mae afiechydon prin yn cynnwys canserau prin fel canserau plentyndod a rhai cyflyrau adnabyddus eraill, fel ffeibrosis systig a chlefyd Huntington.

Cyflyrau heb ddiagnosis

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 DNA y plentyn sydd wedi achosi'r cyflwr. Weithiau mae hyn oherwydd bod y newid mor brin na welwyd mohono erioed o'r blaen ac felly nad oes prawf amdano ym mhrofion arferol clinigau'r GIG. Ar adegau eraill, efallai bod newid yn cael ei weld yn nilyniant DNA y plentyn neu'r person ifanc ond nad oes modd cadarnhau ai hwn yw'r newid sydd wedi achosi'r cyflwr. Mae'r newidiadau hyn yn cael eu galw yn 'amrywiadau na wyddom beth yw harwyddocâd (VUS)'.

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. I blant sydd â syndrom heb enw, efallai bod ganddynt oediad datblygiadol cyffredinol neu'n methu ffynnu. Efallai bod ganddynt anableddau dysgu ac/neu anableddau corfforol.

Weithiau, mae ganddynt anghenion meddygol cymhleth ac efallai epilepsi. Efallai na fydd gan rai plant unrhyw anawsterau dysgu ond efallai y bydd ganddynt anabledd corfforol tra bydd gan eraill anableddau dysgu ond dim anableddau corfforol.

'Pan gawsom y diagnosis, nid oedd ein genetegydd yn gwybod llawer am y cyflwr gan mai dim ond 65 achos sydd yna ar draws y byd.'
Aelod gweithdy

'Fe fuom ni'n ymladd am ddiagnosis i allu cael mynd at y gwasanaethau a'r timau ymyrraeth gynnar a fyddai'n cefnogi fy merch i fod y gorau y gallai hi fod.'
Aelod gweithdy

Rare conditions

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.

1 in 17 people, or seven percent of the population, will be affected by a rare condition at some point in their lives. This equates to approximately 220,000 people in Wales. Most rare conditions are also genetic conditions (so the result of a change in an individual's DNA sequence).

Rare diseases include rare cancers such as childhood cancers and some other well known conditions, such as cystic fibrosis and Huntington's disease.

Undiagnosed 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 their DNA that has caused it. Sometimes this is because the change is so rare it has never been seen before and so isn't tested for in the usual tests available in NHS clinics. On other occasions, a change may be found in the child or young person's DNA sequence but it is not possible to confirm if this is the change that has caused their condition. These changes are called 'variants of unknown significance' (VUS).

Sometimes undiagnosed genetic 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. Some children affected by a syndrome without a name might be described as having global developmental delay or failure to thrive. They might have learning disabilities and/or physical disabilities.

They can sometimes have complex medical needs and may have epilepsy. Some children may not have any learning difficulties but may be physically disabled whilst others are physically unaffected but have learning disabilities.

'When we were given the diagnosis our geneticist didn't know much about the condition as there are only 65 known cases worldwide.'
Workshop participant

'We fought for a diagnosis to be able to access the services and early intervention teams that would support my daughter to be the best version of herself she could be.'
Workshop participant

1.2 Beth mae'n ei olygu i gael cyflwr genetig, prin neu heb ddiagnosis?

1.2 What does it mean to have a genetic, rare or undiagnosed condition?

Gall yr effaith o gael cyflwr genetig, prin neu heb ddiagnosis amrywio'n fawr a gall effeithio ar lawer o agweddau ar fywyd teuluol.

The effects of having a genetic, rare or undiagnosed condition can be very varied and can affect many aspects of family life.

Bydd y ffordd y mae'r cyflwr yn effeithio ar eich teulu 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
- gofal wedi'i drefnu'n wael, gweld llawer o wahanol weithwyr proffesiynol gofal iechyd a llawer o apwyntiadau
- trafferth cael gwybodaeth a chymorth gan arbenigwr, neu grŵp i gleifion, ar gyfer cyflwr eu plentyn.

Gallwch ddarganfod mwy am sut i gael diagnosis ar gyfer cyflerau genetig, prin neu heb ddiagnosis yn **2 Adnoddau Prin: Y daith at ddiagnosis**

How it affects your family will depend on a number of factors but it is not uncommon for families of children and young people with genetic, rare and undiagnosed conditions to experience similar challenges.

These often include:

- A long journey to diagnosis.
- Uncertainty about the future.
- Poorly coordinated care, lots of different healthcare professionals involved and lots of appointments.
- Difficulties in accessing information and support from an expert, or patient group, for their child's condition.

You can find out more about how to obtain a diagnosis for a genetic, rare or undiagnosed condition in **2 Rare Resources: The journey to diagnosis** guide.

Mwy o wybodaeth am gyflyrau genetig, prin a heb ddiagnosis

Further information about genetic, rare and undiagnosed conditions

Mwy o wybodaeth am gyflyrau genetig, prin a heb ddiagnosis:

Genetic Alliance UK
bit.ly/rrgeneticallianceuk

Rare Disease UK
bit.ly/rrrarediseaseuk

SWAN UK (syndromes without a name)
bit.ly/rrswanuk

Your Genome
bit.ly/rryourgenome

Contact a Family
bit.ly/rrcontactmedicalinformation

Orphanet
bit.ly/rrorphanet

Etifeddiad dominyddol
bit.ly/rrdominantinheritance

Etifeddiad enciliol
bit.ly/rrrecessiveinheritance

Etifeddiad cysylltiedig ag X
bit.ly/rrxlinkedinheritance

Further information about genetic, rare and undiagnosed conditions:

Genetic Alliance UK
bit.ly/rrgeneticallianceuk

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SWAN UK (syndromes without a name)
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Orphanet
bit.ly/rrorphanet

Dominant inheritance
bit.ly/rrdominantinheritance

Recessive inheritance
bit.ly/rrrecessiveinheritance

X-linked inheritance
bit.ly/rrxlinkedinheritance

Eich **Nodiadau**

Your **Notes**


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