Language and terminology

This guide, developed by the Participant Panel, recommends how to talk about the people whose data is curated at Genomics England.

Genomics England

Contents

Principles

This guide has been developed following a Participant Panel workshop on 8th February 2022. It sets out the Panel's agreed recommendations on how to talk about the people whose data is curated by Genomics England. This guide is intended for use by Genomics England staff and their partner organisations across the wider genomics ecosystem.

In drawing together our recommendations, the Participant Panel has agreed a set of general principles.

Every data point has a face

Behind each data point is a person; someone who could be just like you. Everyone could be affected by a rare health condition. Rare diseases are collectively so common (1 in 17 people) that everybody will know someone who has one, although they might not talk about it. And cancer will touch most families during their lifetime (1 in 2 people).

So…

- When thinking about communicating with patients and research participants, treat everyone as you would like to be treated. Be mindful of the audience (with the cognition of an 'intelligent 7 year-old') and use accessible formats and accessible language.
- Respect everyone equally, regardless of how they got involved in genomics (patient, family member, professional, etc): it's a team endeavour and we are all equally invested in its success.
- Recognise that patients and families do not always choose to define themselves or their relatives by the health conditions that brought them here: genomics research is just one aspect of their lives.
- When writing up case studies, picture the people whose data you're talking about. Would they be happy with how you are describing them? (See Table 1 on page 6 for further advice).
- Always keep in mind that human flourishing is for all: everyone has something to contribute.
- However, don't be afraid of saying the wrong thing; it's always better to come and talk to patients and research participants, than to avoid it for fear of causing offence!

Drivers and differences

Those who agree to donate their data have many different reasons for doing so:

- The 100,000 Genomes Project recruited two groups: those with undiagnosed rare conditions and those with cancer.
    - 'Rare disease' participants signed up for diagnoses, better understanding of their conditions now and in the future, and possible treatments to alleviate their effects. Few were anticipating cures. Getting a long-awaited diagnosis creates choices about what to do with that information, where to look for treatment and also means people can link with others with the same condition, on the way to understanding it better.
    - Cancer participants had different perspectives: those who signed up already knew that they had cancer, and understood that the Project could not return genetic information in time to help them personally. Their reasons for involvement were principally altruistic.
- As WGS becomes available through the NHS Genomic Medicine Service, it is generating a set of perspectives and drivers which may be different to recruits to the 100,000 Genomes Project, e.g.
    - 'For rare disease' participants these may now include meeting an immediate clinical need or the opportunity to find out more about their conditions in due course.
    - Cancer patients on the other hand, now that return of results is so much quicker, are being offered the chance to look for actionable variants, which may be their main driver.

For example, those who were recruited to the 100,000 Genomes Project from the Deaf community, many don't see themselves as disabled, and would not necessarily want this to change.

Participants are not a homogenous group. They have different approaches / cultures / language fluency: don't assume that 'one size fits all' in your communication with participant communities.

Know your audience: family journeys bring different stages of acceptance / grief / etc – newly diagnosed families may feel very differently to those who have been living with a condition for longer. Generally speaking, acceptance grows with time since diagnosis. But so does an awareness of stereotyping and the disabling nature of many aspects of mainstream society: the 'social model of disability'.

Use of language

We all recognise that the same term can evoke different reactions, but there are some that should generally be avoided (see Table 1 on page 7 for details). Whilst it is understood that clinicians talking to colleagues will use scientific language amongst themselves, they should be aware that increasingly their notes are shared with patients and to be mindful of their impact that scientific language may have in other contexts.

Bear the context in mind when deciding what language to use when describing other people's experiences: a specific diagnosis may indeed have been 'devastating' for the particular family in question, but the condition itself may not be so for everybody. If you have any means of checking with the participant/patient/family that you wish to present in a case study, please do ask them how they actually feel (or felt when the incident occurred) before speaking on their behalf.

Talk to people directly affected or living with a condition, and present some positive experiences when writing information for patients and parents. Medical textbooks rarely reflect the real lived experiences of people with particular conditions, by typically using a uniformly negative tone. Clinicians often repeat this same language in patient literature. This is especially alarming for newly diagnosed patients and families, who are on a voyage of discovery that they may never have anticipated but will have life-long implications.

Disabled people may choose terms to describe themselves and their own communities that are not acceptable labels when applied by outsiders; when starting to work with a particular community, take time to ascertain the terminology they'd like you / professionals / outsiders to use.

Avoid making assumptions, and using value-laden descriptions of life experience such as 'suffering', 'victims' to be 'pitied' etc.

If you must talk about the impact of a condition, strive to use language that describes the symptoms in an objective way (e.g. the patient's condition results in decreased hand-eye co-ordination).

Avoid sensationalising disabled people, cancer patients or their families, e.g. suggesting they're a 'superhero' or 'brave' or 'inspirational' for living with these conditions. You'd probably do the same if you were in their position.

Avoid inferring or implying things about how people look, move or behave based on their DNA. Always be clear whether you are talking about the person or their unusual gene(s). Terms like 'mutation' are OK to use when talking about genes, but not OK when talking about a person. Please see Table 2 on page 7 for details.

Fluidity

We all recognise that acceptable uses of language evolve over time. Keep refreshing your understanding of acceptable language e.g. regularly seeking input from experts / people with lived experience of the conditions that you are working on.

Active learning: what does your audience think about what has been presented? Don't be afraid to ask.

Please contact the Panel at any time and talk to us about language – we welcome these conversations and are happy to share our lived experiences – that's why we are here!

Talking about disability – recommendations

If you're tempted to use…	Go for this instead please
Victim of	Person who has / Person with / Person who experienced
Crippled by	Disabled person / Person who has / Person with
Sufferer Suffering from	Person who has Person with / Affected by…? / Living with?
Afflicted Afflicted by	Person who has Person with
Wheelchair bound	Wheelchair user. Wheelchair users often see them positively: they offer freedom and independence
Invalid	Disabled person
Handicap	Disability / with an impairment
Handicapped person/ Person with a disability	Disabled person
Disability	Condition / impairment
Less-abled/differently-abled	Disabled people
Able-bodied	Non disabled people
The disabled	Disabled people
The blind	Blind person / people / Visually impaired
The deaf	Deaf person / people
Deaf and dumb Deaf mute	Deaf or deafened Hearing impaired
Disfigured	Facial disfigurement is the preferred term. No need for adjectives (severe / mild etc)
Missing limb	Limb difference
Person with autism	Autistic person (but see also NHSD guidance re: epilepsy, diabetes etc which suggests 'a person with x' rather than 'an epileptic' or 'a diabetic')
Mental handicap	Learning difficulty or learning disability, depending on the severity of the condition
Mute / dumb	Speech difficulty, non-verbal, preverbal
Mad / insane	Experiencing mental ill health
Mentally ill	Experiencing mental ill health
Dwarf / Midget	Short person / Short stature
Deformity / visible difference	Disfigurement (no adjectives necessary: avoid mild / severe etc)
Congenital (condition)	Health condition present from birth

Talking about genomics – recommendations

If you're tempted to use…	Go for this instead please
(Germline) mutation …when referring to the DNA we're born with	Preferred: (gene) variant Possible alternatives: (gene) change, spelling mistake, alteration, glitch
(Somatic) mutation … when used to describe changes that occur in cells throughout life, cancer, etc	Mutation or variant is OK
Dysmorphic	OK in clinical / technical context NOT OK when talking to patients and families – e.g. instead describe features as 'unusual'
Defective (system in the body)	OK in technical context, some patients also happy with this
Genetic defect or fault (in a person)	Please avoid. Prefer the technical language (e.g. deletion, insertion, duplication, variant)
Disease	Condition or Disorder (especially if part of the name of the diagnosis e.g. OCD)
Normal	Typical (e.g. neurotypical) Or 'born without [the particular condition/variant]', 'born after an uneventful pregnancy'
Abnormality	Unusual / atypical (e.g. neurodiverse) Or 'born with [the particular condition/variant]'
Devastating, terrible, etc. (disease)	Avoid making value judgements about other people's experiences
'Fixing' people	Finding treatments for people [that could improve their quality of life]
Rare disease	Rare condition 'rare disease' is commonly used, but many of us prefer 'rare condition'.
Rare syndrome	OK in medical / technical context
Exploiting data Interrogating data Harvesting data Mining data	Accessing, using or analysing data
Healthy relatives (compared to proband)	(Unaffected) relatives or relatives without the condition, symptom or gene variant in question

How to talk about cancer

- Don't use war metaphors! Battling, fighting, winning etc suggest that individuals can 'overcome' it by force of will, which is just not true.

- Do recognise that Genomics England's cancer cohort is made up of people with a spectrum of experiences of cancer, not all of whom are patients. They include:

    - People living with cancer
    - People who have had cancer (avoid 'cured' or 'survivor')
    - People with a cancer susceptibility gene variant (avoid 'previvor')

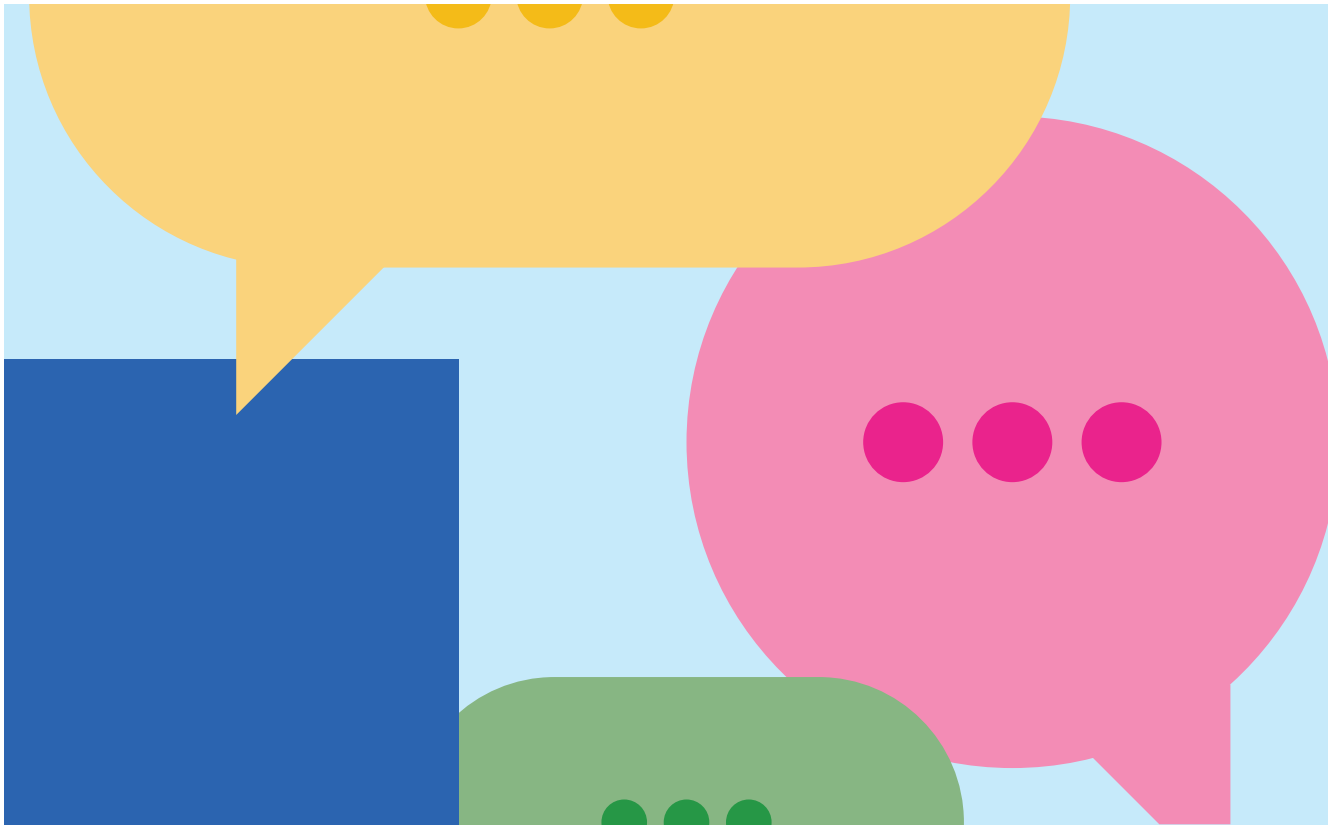
How to talk about the Participant Panel

The Participant Panel is an independent entity. It should be described as:

- the [independent] Participant Panel at Genomics England, or
- the Participant Panel, or
- the Panel.

It is not 'the Patient Participant Panel'.
It is not 'Genomics England's Participant Panel'.

It is not a UK Participant Panel – it represents the people whose data is in the 'National Genomics Research Library' but some of the devolved nations have their own arrangements.



How to talk about the 100,000 Genomes Project and its participants

- Note that it's genomes in the plural and that Genomes and Project should always be capitalised. 100kGP is acceptable on slides and internal documents. Externally it's the 100,000 Genomes Project.

- We always talk about 'participants' not patients. This is because:

    - 'Participant' correctly implies active involvement and choice.
    - At least two thirds of our rare disease cohort are not patients – they are unaffected relatives who do not have the same condition or genetic variant
    - Many people with rare conditions do not consider themselves as 'patients' which implies that they are unwell. For example, they may have hearing loss.
    - Many people previously treated for cancer or who are at risk from familial cancer syndromes do not consider themselves as 'patients'.

- Please do not describe the 100,000 Genomes Project as 'finished', 'completed' or 'done'. Between 75-80% of those recruited to the 100,000 Genomes Project due to a rare disease/ condition have NOT had a diagnosis, although the Project has completed its recruitment and initial return phase, the quest for diagnosis continues for them and needs to remain a focus for Genomics England.