Breakout session -
WG10 Common and Complex Disease
WG12 - Genome of Europe

Chair: Andres Metspalu

Co-chair: Serena Scollen

04

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WG10 and WG12

To talk about personalised medicine and prevention concept into health care, which is based on genetic information of the individuals, **we have to understand the genetic variation between the people and use it accordingly**.

Human genetics and genomics research over the last 10-15 years provided enough information for it.

DNA sequencing and genotyping costs have reached the point that now allows population based approaches, which is needed in secondary personalised prevention, economically feasible.

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WG10 plan

1. Genotyping up to 1-5% population-based samples using high density SNP microarrays with imputation against the population specific WGS reference database (generated by WG12 / Genome of Europe)
2. Developing or implementing an existing platform for decision support systems for Polygenic Risk Scores (PRS) - specific algorithms that in addition to genomic (-omics) risks can include environmental (e.g.smoking) and lifestyle risks e.g. BMI to determine personal risk for common complex disorders
3. Construction of Polygenic Risk Scores (PRS) of the most common diseases based on the data from large GWAS studies and for likely drug response (using CPIC as a reference) for 30-50 mostly used prescription drugs based on 10-15 pharmacogenetically important gene variants, and clinical validation
4. Identifying people with high disease risk and establishing large scale population-based early intervention programs for the prevention of various common and complex diseases. At the beginning the existing screening programs (mammography in breast cancer screening, CRC, cervical cancer, CVD etc., T2D) could be modified accordingly.

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WG10: What is needed from policy for responsible implementation?

A) Public and professional trust: ensuring that the general public, researchers, clinical professionals and policy makers are well informed about genomics and feel empowered to make decisions, in order to ensure its uptake by (public) healthcare systems and integration into personalised healthcare

B) Focus on inclusion and equity: avoiding an increase in unequal access to health services

C) Sequencing facilities and personnel: ensuring that facilities and workforce are facilitated for sequencing, but also counselling and support

D) IT-infrastructure: a) ensuring that appropriate technical infrastructure is available, allowing for secure, federated access to genomic data, b) implementing interoperability guidelines to achieve data of internationally agreed standards

E) Legal framework allowing safe data-exchange and informed choice:safeguarding privacy and personal control

F) Financial arrangements/reimbursement of testing: ensuring access to testing for citizens

G) Piloting implementation: assessing evidence and identifying and overcoming practical and remaining ethical, legal and societal issues

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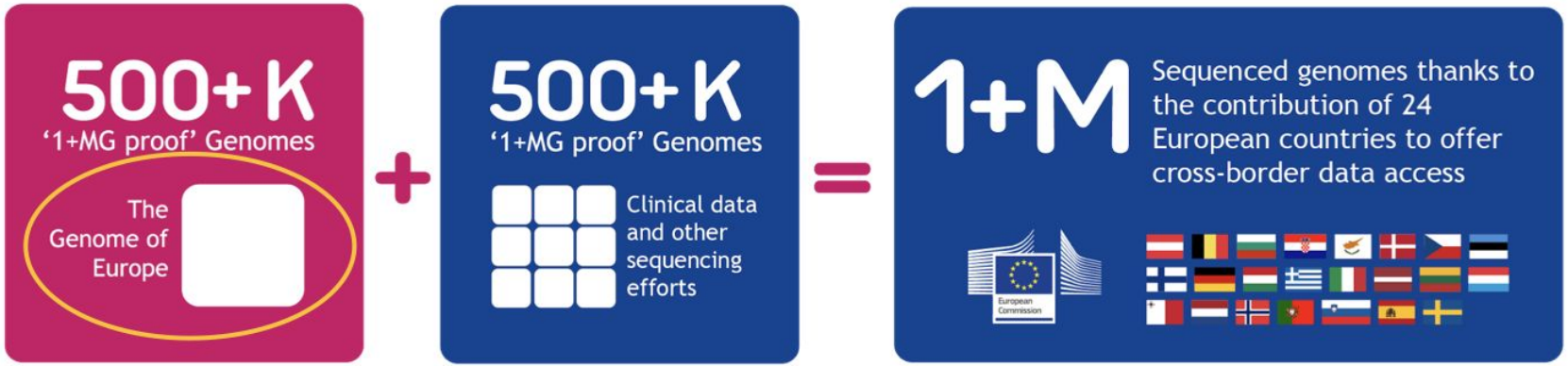
WG12: Genome of Europe

European commitment to achieve access to at least 1 million sequenced genomes by 2022

B1MG is creating legal guidance, best practices, recommendations and an infrastructure to achieve the goal



- Minimal phenotype: age, sex, migration background (ancestry)
- Special attention to include minorities
- 38 ancestries included
- 1,000 genomes as a minimal sample size per ancestry group and/or per country
- Gathering the first WGS from countries in the next 6 months

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Feedback

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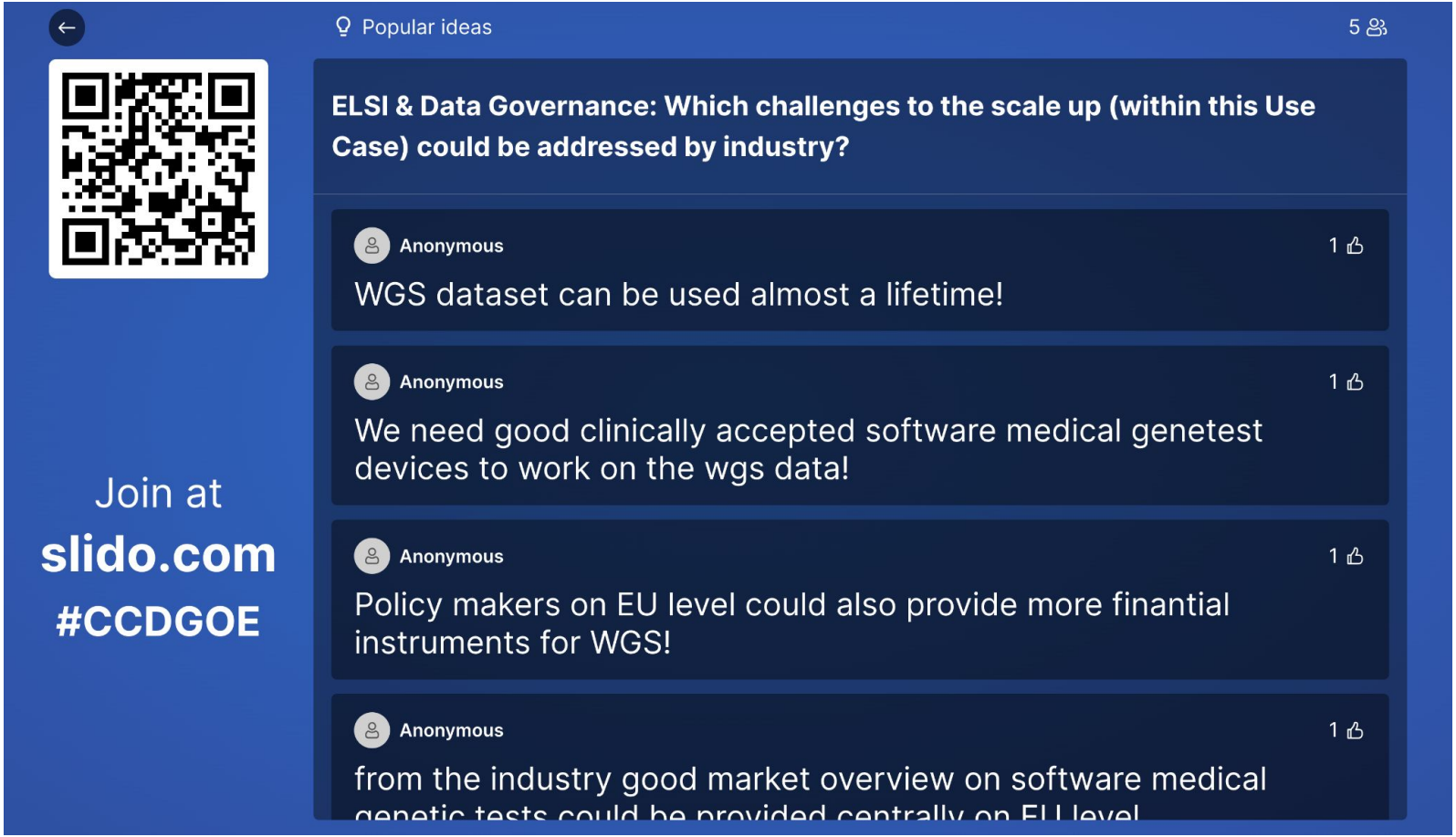
Popular ideas
5
ELSI & Data Governance: Which challenges to the scale up (within this Use Case) could be addressed by industry?
Anonymous
1
WGS dataset can be used almost a lifetime!
Anonymous
1
We need good clinically accepted software medical genetest devices to work on the wgs data!
Anonymous
1
Policy makers on EU level could also provide more finantial instruments for WGS!
Anonymous
1
from the industry good market overview on software medical
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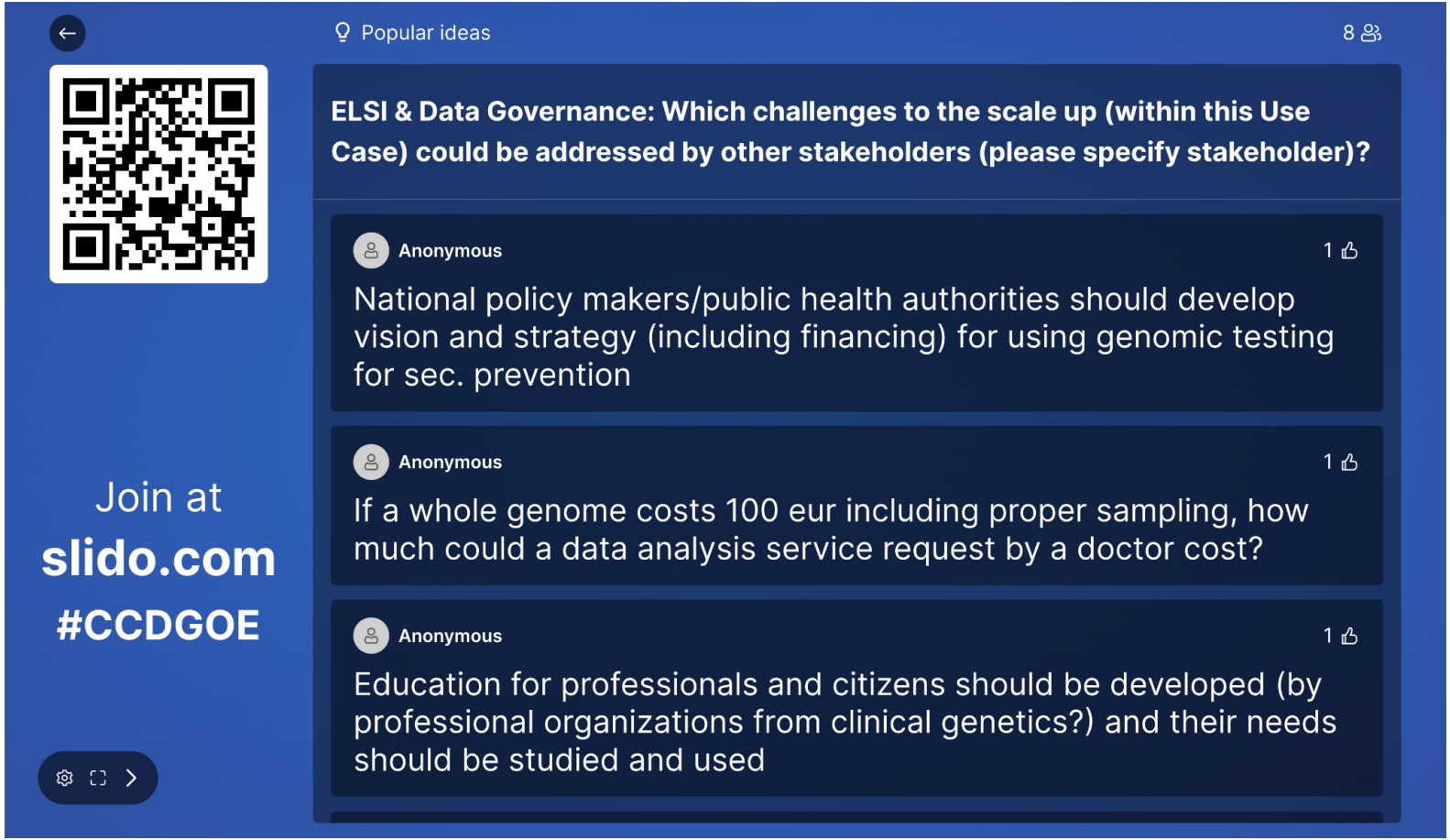
Popular ideas
8
ELSI & Data Governance: Which challenges to the scale up (within this Use Case) could be addressed by other stakeholders (please specify stakeholder)?
Anonymous
1
National policy makers/public health authorities should develop vision and strategy (including financing) for using genomic testing for sec. prevention
Anonymous
1
If a whole genome costs 100 eur including proper sampling, how much could a data analysis service request by a doctor cost?
Anonymous
1
Education for professionals and citizens should be developed (by professional organizations from clinical genetics?) and their needs should be studied and used
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Popular ideas 3

Data & Quality Standards: Which challenges to the scale up (within this Use Case) could be addressed by industry?

Anonymous 1

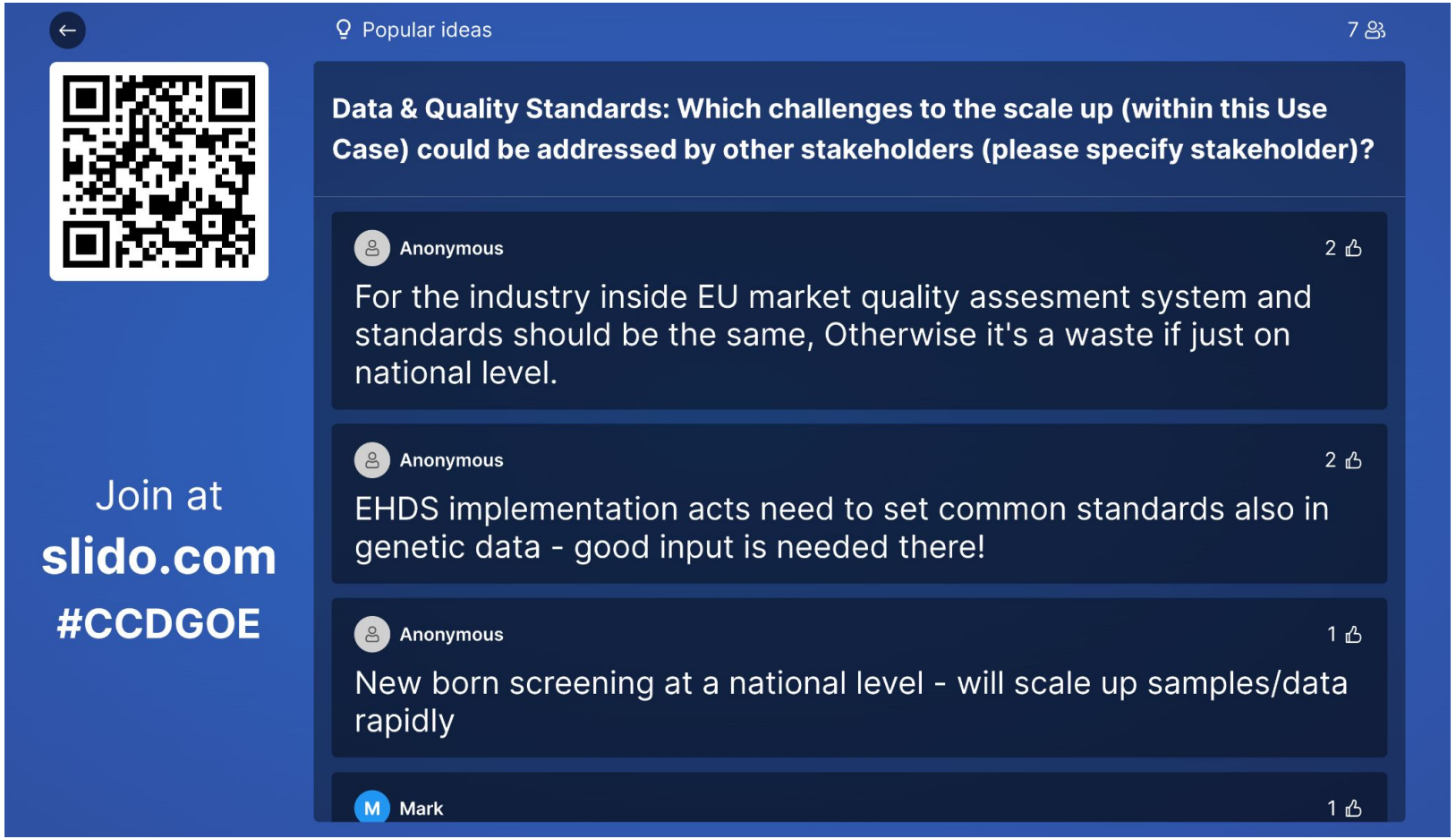
Healthcare - need to set the quality etc; regulation to be set at a government level

Anonymous 0

inter lab quality assessment - industry can produce chips etc that respond to the quality control set

M Mark 0

I may have missed this example earlier but Screen4Care is EFPIA supported on newborn screening using genomics. Could this be extended


Popular ideas
7
Data & Quality Standards: Which challenges to the scale up (within this Use Case) could be addressed by other stakeholders (please specify stakeholder)?
Anonymous
2
For the industry inside EU market quality assesment system and standards should be the same, Otherwise it's a waste if just on national level.
Anonymous
2
EHDS implementation acts need to set common standards also in genetic data - good input is needed there!
Anonymous
1
New born screening at a national level - will scale up samples/data rapidly
M
Mark
1
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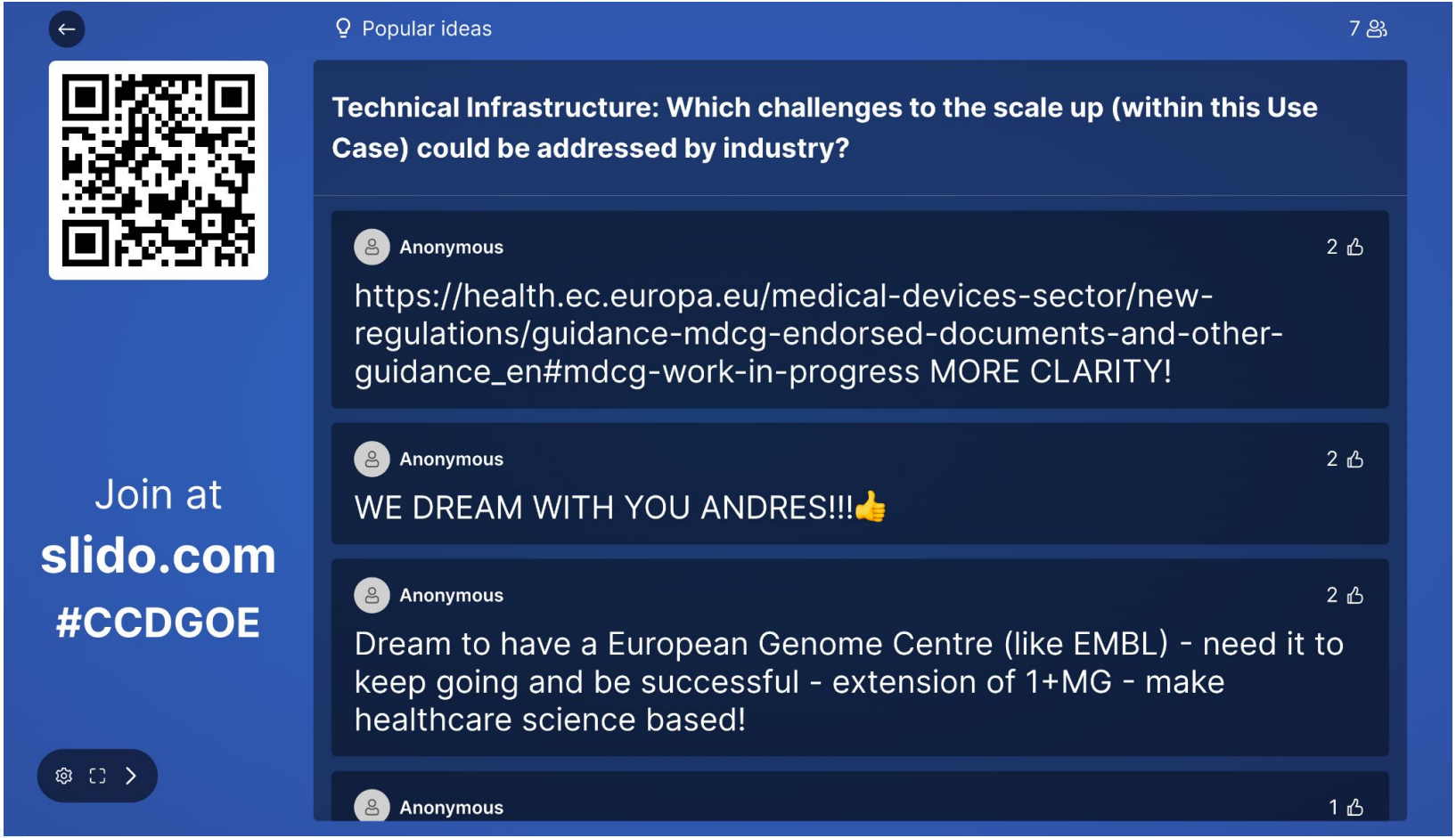
Popular ideas
7
Technical Infrastructure: Which challenges to the scale up (within this Use Case) could be addressed by industry?
Anonymous
2
https://health.ec.europa.eu/medical-devices-sector/new-regulations/guidance-mdcg-endorsed-documents-and-other-guidance_en#mdcg-work-in-progress MORE CLARITY!
Anonymous
2
WE DREAM WITH YOU ANDRES!!!👍
Anonymous
2
Dream to have a European Genome Centre (like EMBL) - need it to keep going and be successful - extension of 1+MG - make healthcare science based!
Anonymous
1
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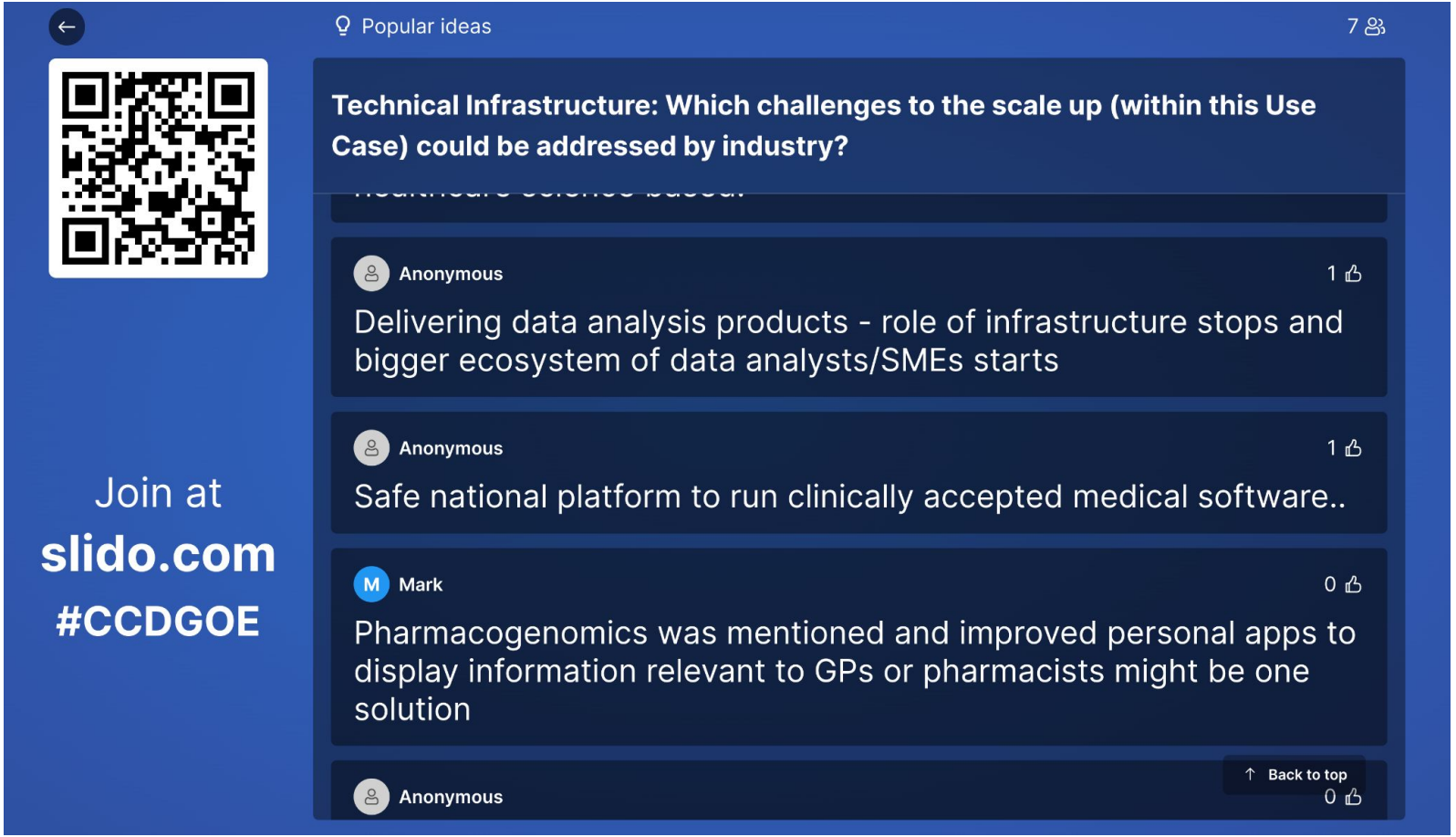
Popular ideas
7
Technical Infrastructure: Which challenges to the scale up (within this Use Case) could be addressed by industry?
Anonymous
1
Delivering data analysis products - role of infrastructure stops and bigger ecosystem of data analysts/SMEs starts
Anonymous
1
Safe national platform to run clinically accepted medical software..
M
Mark
0
Pharmacogenomics was mentioned and improved personal apps to display information relevant to GPs or pharmacists might be one solution
Back to top
Anonymous
0
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‘Ideas’ question 7

Thank you!

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