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Implementing Genomic Medicine: An Evaluation of the Hong Kong Genome Project (HKGP)

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Declaration of Interest

Funding Body:
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Key highlights

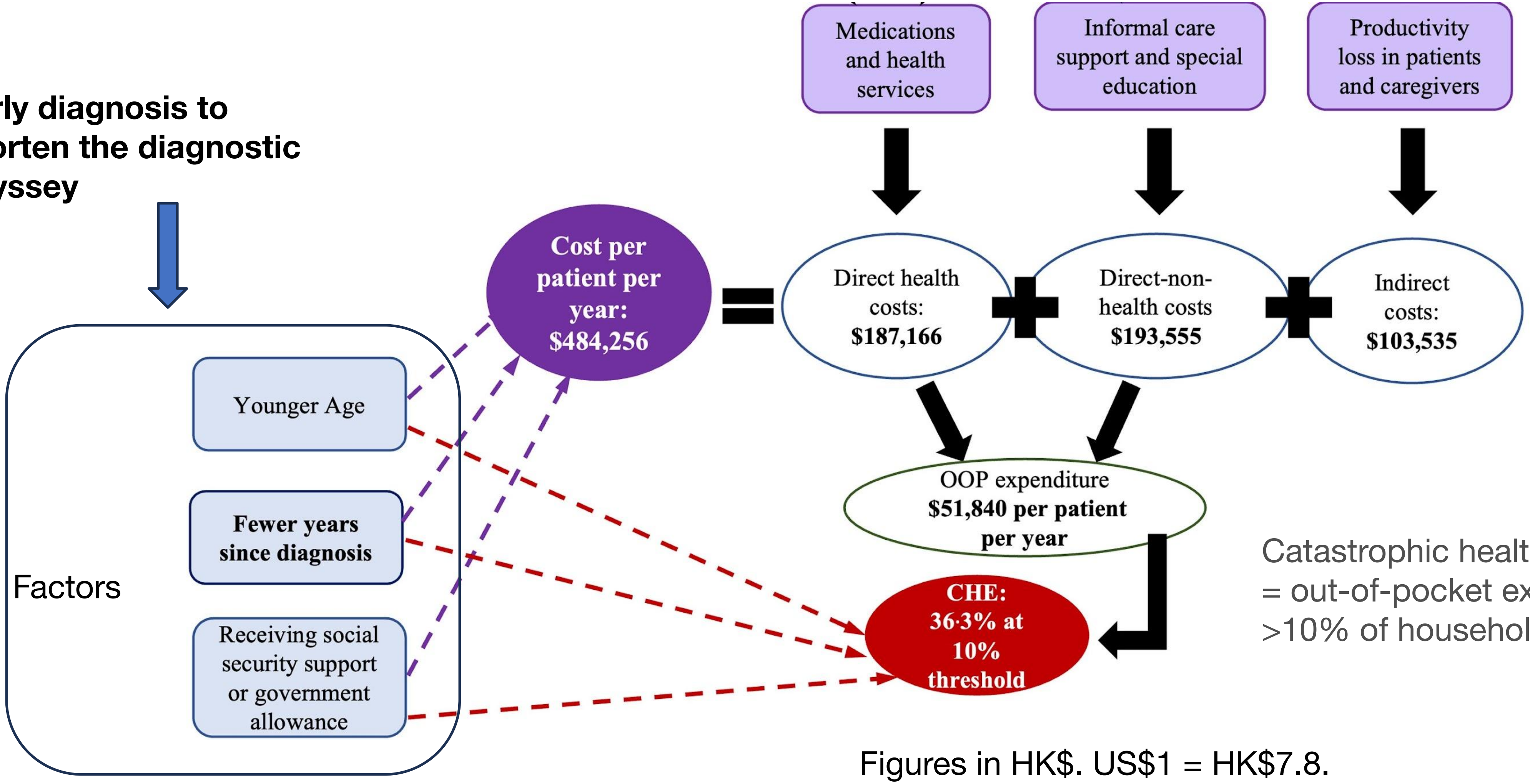
- Integration of genomic medicine into healthcare system could be facilitated by advancing scientific research, nurturing talents, and enhancing genomic literacy in general population
- Participants generally satisfied with the earlier phases of HKGP
- Future research warranted for clinical validation, cost-effectiveness and psycho-social-economical outcomes

CDC’s framework for program evaluation

1. Engage stakeholders
2. Describe the program
3. Design evaluation methods
4. Gather credible evidence
5. Justify conclusions
6. Ensure issues addressed and share lessons learned

Socio-economic costs for rare diseases in Hong Kong

Early diagnosis to shorten the diagnostic odyssey



Medications and health services

Informal care support and special education

Productivity loss in patients and caregivers

Cost per patient per year: $484,256

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Direct health costs: **$187,166**

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Direct-non-health costs **$193,555**

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Indirect costs: **$103,535**

OOP expenditure **$51,840 per patient per year**

Factors

Younger Age

Fewer years since diagnosis

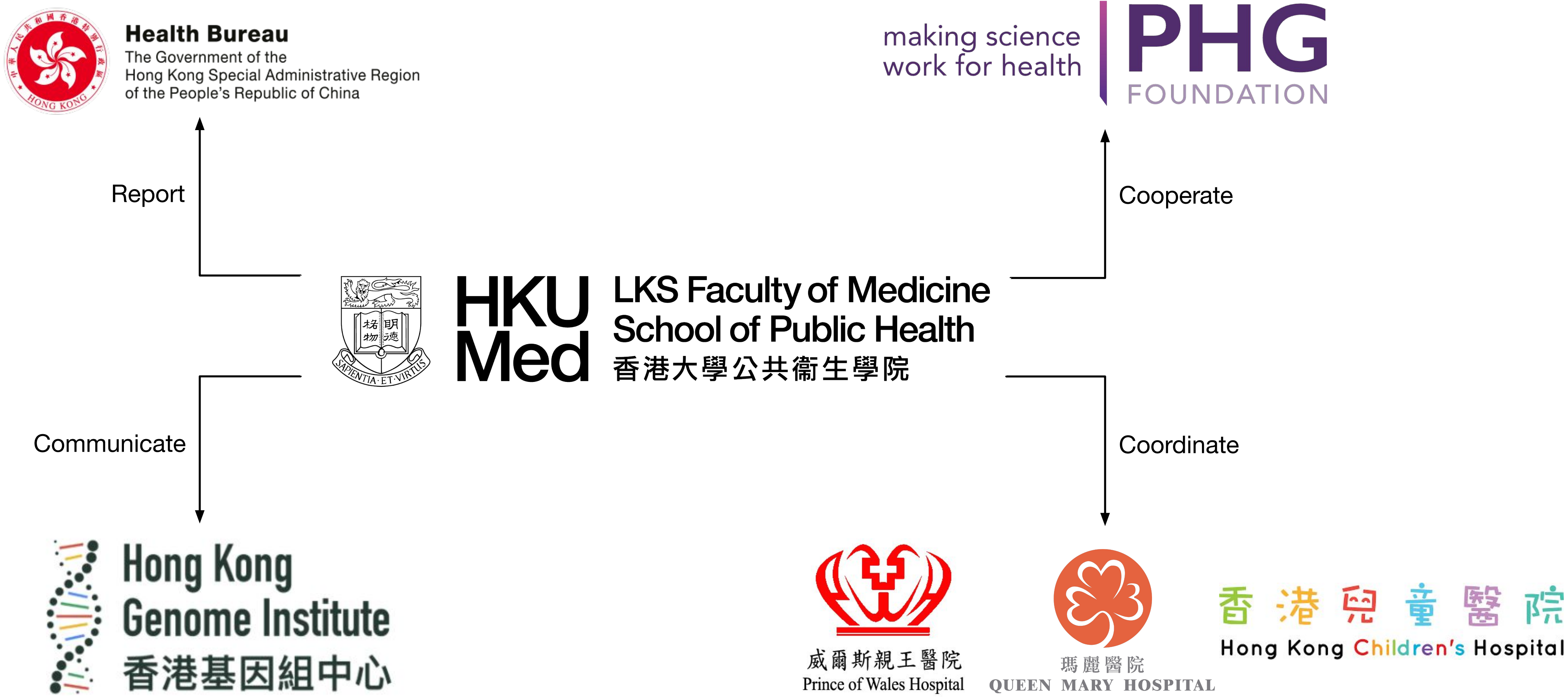
Receiving social security support or government allowance

CHE: 36·3% at 10% threshold

Catastrophic health expenditure = out-of-pocket expenditure >10% of household income

Figures in HK$. US$1 = HK$7.8.
(Chung et al., 2023 doi:10.1016/j.lanwpc.2023.100711)

Establish ties with stakeholders



Commissioning body

Health Bureau
The Government of the
Hong Kong Special Administrative Region
of the People's Republic of China

Report

Health policy consultant

making science work for health
PHG FOUNDATION

Cooperate

HKU Med
LKS Faculty of Medicine
School of Public Health
香港大學公共衛生學院

Communicate

Hong Kong Genome Institute
香港基因組中心

Implementing agent of HKGP

Coordinate

威爾斯親王醫院
Prince of Wales Hospital

瑪麗醫院
QUEEN MARY HOSPITAL

香港兒童醫院
Hong Kong Children's Hospital

Provides resources for project execution

Hong Kong Genome Project (HKGP)

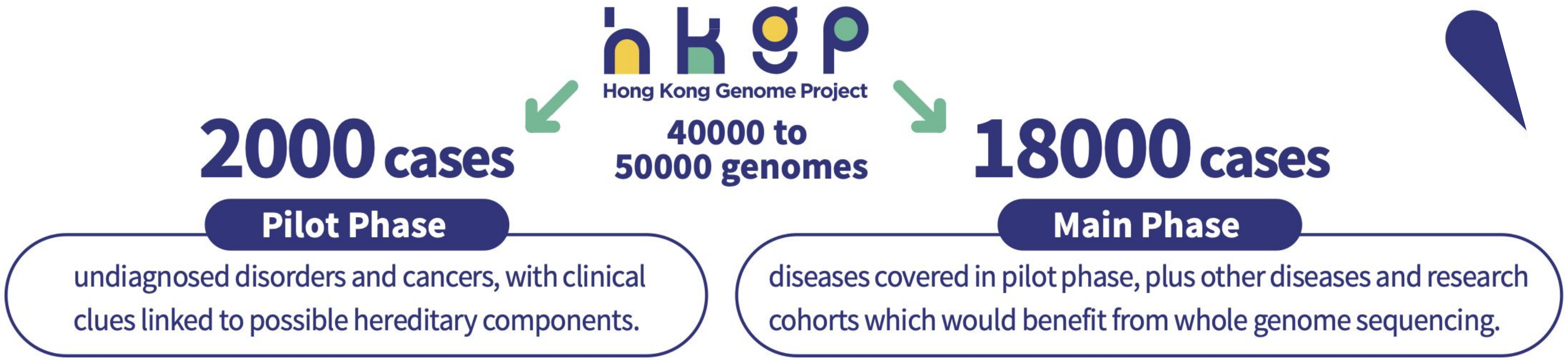
2017
Government announced formation of steering committee

2018
Government announced that HKGP is under way

2020
HKGI established with key objective to conduct HKGP

2021
HKGI commenced full operation
HKGP launched

2022
SPH embarked on evaluation project of HKGP



40000 to 50000 genomes

2000 cases

Pilot Phase

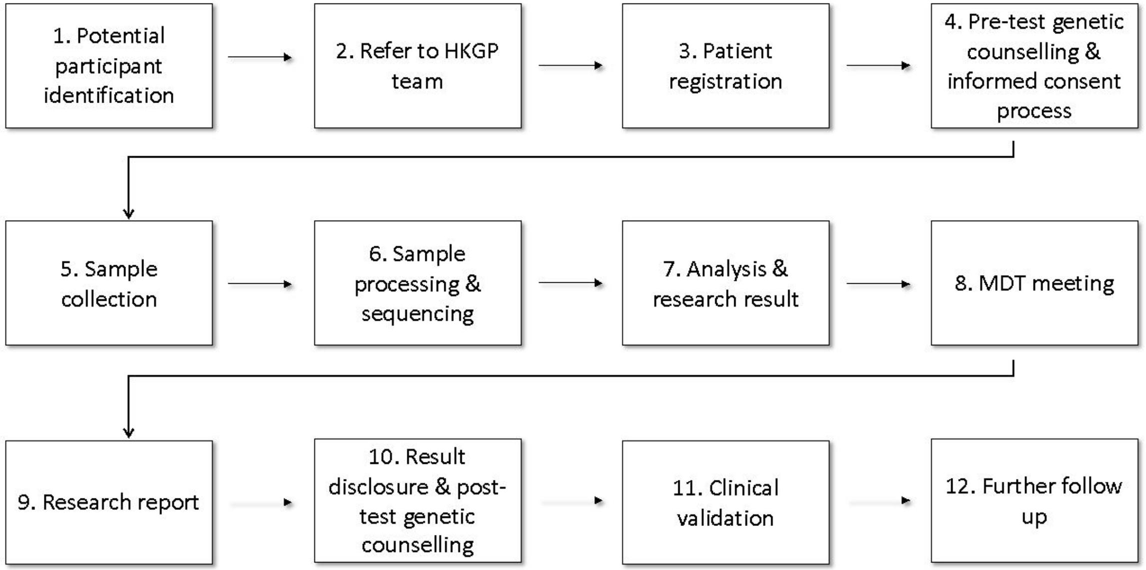
undiagnosed disorders and cancers, with clinical clues linked to possible hereditary components.

18000 cases

Main Phase

diseases covered in pilot phase, plus other diseases and research cohorts which would benefit from whole genome sequencing.

Workflow


1. Potential participant identification
2. Refer to HKGP team
3. Patient registration
4. Pre-test genetic counselling & informed consent process
5. Sample collection
6. Sample processing & sequencing
7. Analysis & research result
8. MDT meeting
9. Research report
10. Result disclosure & post-test genetic counselling
11. Clinical validation
12. Further follow up

Enrollment: By invitation, free-of-charge

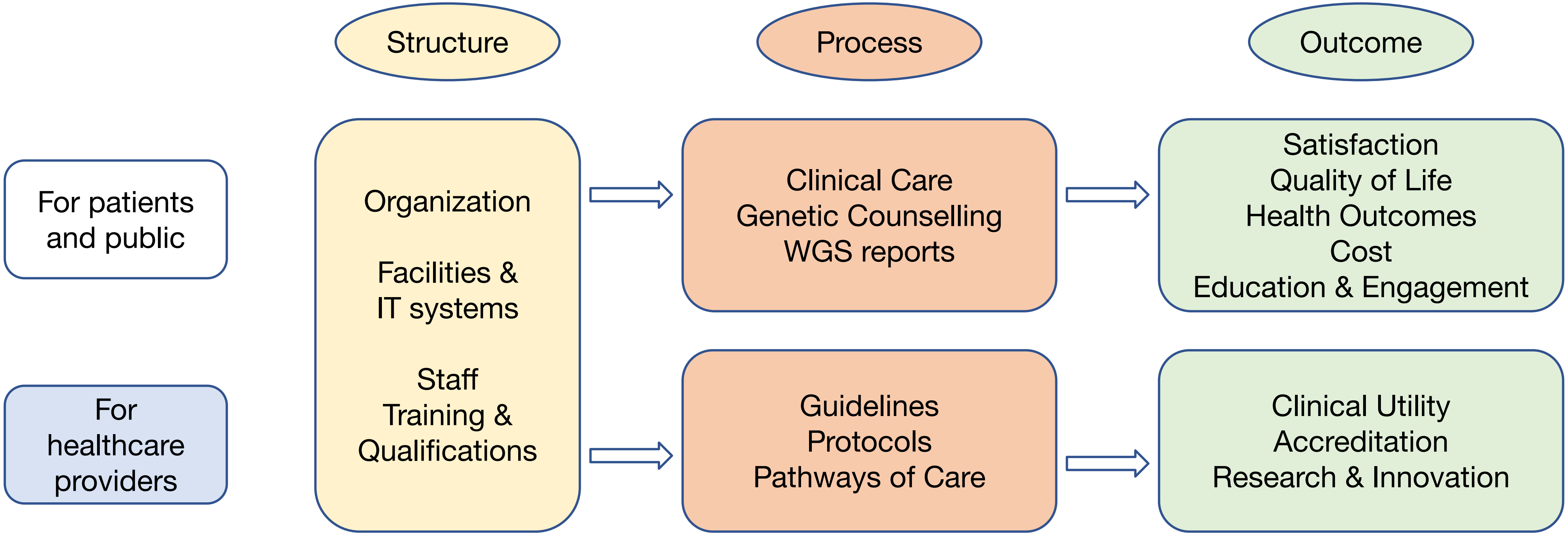
Partner Centers: PWH, QMH, HKCH

Eligibility: Patients with
1) undiagnosed diseases or
2) hereditary cancers

Some participants of **HKGP** were then invited to also enroll in the **evaluation project**

Methods: Mixed methods (Survey □ Focus group)

Donabedian approach for evaluation



Structure

Process

Outcome

For patients and public

For healthcare providers

Organization

Facilities & IT systems

Staff Training & Qualifications

Clinical Care
Genetic Counselling
WGS reports

Guidelines
Protocols
Pathways of Care

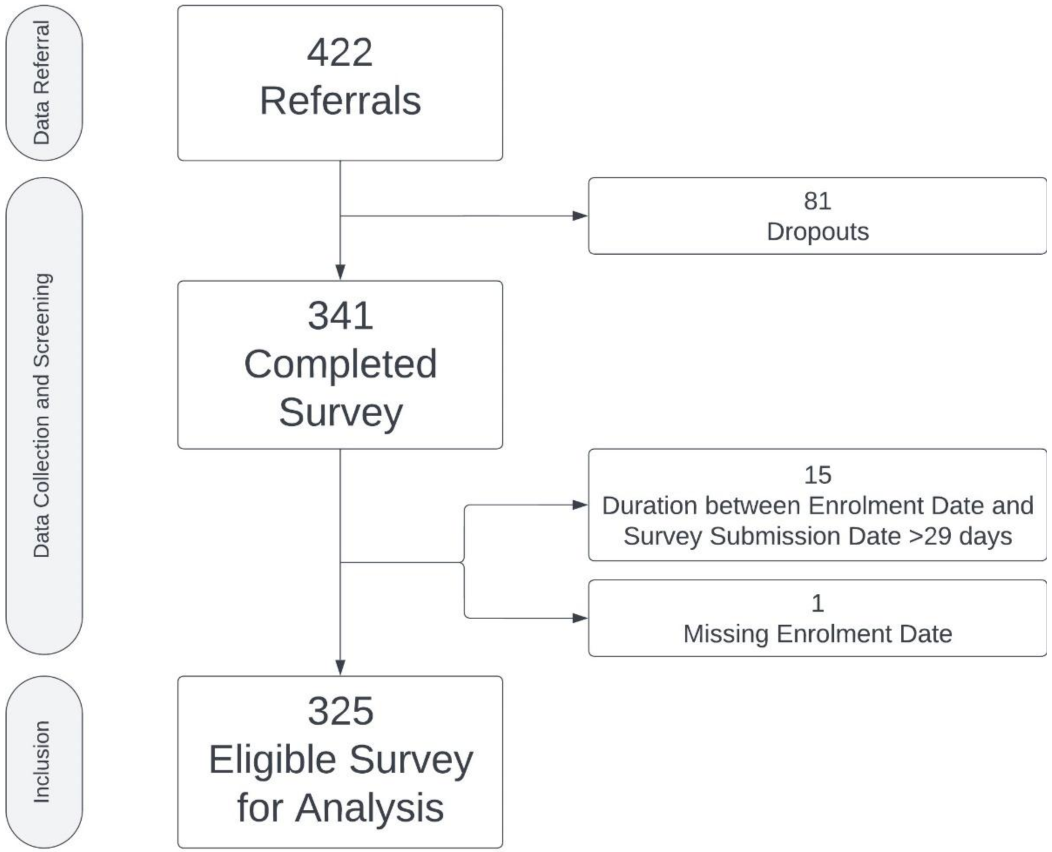
Satisfaction
Quality of Life
Health Outcomes
Cost
Education & Engagement

Clinical Utility
Accreditation
Research & Innovation

Mixed-methods study design

	Quantitative: Population Survey	Qualitative: Focus Group Interview
No. of participants:	>287	>20
Study Procedure	Informed consent by on-site genetic counsellors/ nurses Telephone survey done by SPH staff Response entered and managed on Qualtrics	Online Zoom interview by SPH staff
Development of tools	Reviewed existing literature Self-created survey was reviewed for content validity and feasibility	Self-created semi-structured interview guide
Remarks	Only analyze response that were recorded within 30 days of genetic counselling session	Assigned participants into different groups to minimize groupthinking

Overview of results



Data Referral

422
Referrals

81
Dropouts

Data Collection and Screening

341
Completed Survey

15
Duration between Enrolment Date and Survey Submission Date >29 days

1
Missing Enrolment Date

Inclusion

325
Eligible Survey for Analysis

	Quantitative: Population Survey	Qualitative: Focus Group Interview
Study Period:	13 June – 27 Sept 2023	4 Sept – 12 Sept 2023
No. of participants:	325 (422 referrals, response rate: 77%)	21

Demographics

Characteristic	**Survey participants, n = 325 (%)**	**Focus group participants, n= 21 (%)**
Age group, years		
≤19	50 (15.4)	4 (19.0)
20-39	81 (24.9)	7 (33.3)
40-59	105 (32.3)	2 (9.5)
≥60	89 (27.4)	8 (38.1)
Gender		
Male	159 (48.9)	7 (33.3)
Female	165 (50.8)	14 (66.7)
Other	1 (0.3)	0 (0)
Ethnicity		
Chinese	320 (98.5)	21 (100)
Other	5 (1.5)	0 (0)
Marital Status		
Married	196 (60.3)	13 (61.9)
Other Marital Status	129 (39.7)	8 (38.1)
Household Income Level, HK$		
≤$19,999	35 (10.8)	4 (19.0)
$20,000–$29,999	57 (17.5)	1 (4.8)
$30,000–$39,999	38 (11.7)	3 (14.3)
$40,000–$59,999	49 (15.1)	5 (23.5)
≥$60,000	63 (19.4)	4 (19.0)
(Refused to answer)	83 (25.5)	4 (19.0)
Housing		
Public	108 (33.2)	8 (38.1)
Private	177 (54.5)	11 (52.4)
Other	29 (8.9)	0 (0)
(Refused to answer)	11 (3.4)	2 (9.5)
Educational		
Primary or below	58 (17.8)	4 (19.0)
Secondary	135 (41.5)	11 (52.4)
Tertiary	132 (40.6)	6 (28.6)

Findings: Quantitative Survey

Statement	Agree % [95% CI]
Satisfied with overall experience of the Hong Kong Genome Project (HKGP)	89.8 [86.1, 92.7]
Confident that my personal health data will be securely protected.	88.6 [84.7, 91.6]
Genetic counselling session helped me to identify what I needed to know to make decisions about what would happen to me.	84.6 [80.3, 88.1]
Genetic counselling session was helpful to me.	87.1 [83.0, 90.3]
Feel that taking part in HKGP could benefit others.	86.8 [82.7, 90.0]
Feel that taking part in HKGP could advance genomic research in Hong Kong.	88.9 [85.0, 91.9]
Feel that taking part in HKGP could improve my / my child's medical treatments.	73.5 [68.5, 78.0]
Would recommend fellow patients or those in similar needs to participate in the HKGP	79.4 [74.7, 83.4]

Findings: Quantitative Survey

Results on Net Promoter Score
Value: >20 □ Favourable ; >50 □ Excellent

Has confidence to join project due to possibility to withdraw anytime	54
Participation would advance research in HK	53
Confident that personal health data will be securely protected	51
Participation would benefit others	48
Overall experience of HKGP	45
Genetic counselling session was of proper length of time	44
Genetic counselling session was helpful	41
The informed consent session was helpful in understanding project	40

Findings: Quantitative Survey

Participants with secondary and tertiary education were less likely to agree that the counselling was the right length of time (OR: 0.12, 95% CI: 0.015-0.83 and OR: 0.12, 95% CI: 0.13-0.98, respectively)

Less likely to agree the genetic counselling session helped make decisions about themselves (OR: 0.08, 95% CI: 0.01-0.62; and OR: 0.08, 95% CI: 0.01-0.76, respectively)

Also less likely to agree that genetic counselling was helpful (OR: 0.023, 95% CI: 0.001 – 0.32, and OR: 0.023, 95% CI: 0.001-0.40, respectively).

40-59 year olds were more likely to agree that genetic counselling was the right length of time (OR: 7.94, 95% CI: 1.11-60.58),

More likely to agree that genetic counselling helped them to make decisions about themselves (OR: 9.00, 95% CI: 1.15-69.92).

Also much more likely to agree that genetic counselling was helpful (OR: 26.97, 95% CI: 2.35-377.34).

Findings: Focus Group

Reasons for participation	Helping the scientific community and future patients Using results to guide decision-making in family planning Getting confirmatory diagnosis for condition
Shortening diagnostic odyssey	Turnaround time was too long to be practically useful Reduce time for deliberating on therapeutic decision
Overall Perception	All expressed overall satisfaction in study Genetic counselling sessions were clear and comprehensible Illustration of concepts without using obscure scientific jargons

1 Engage Stakeholders
2 Describe the program
3 Design evaluation methods
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6 Put to use and lessons learned

Findings: Focus Group

Reasons not to conduct genetic tests in other settings	Cost Turnaround time Privacy Necessity
Areas for consideration	Optimize workflow to shorten turnaround time Devise better follow-up mechanisms Consider territory-wide implementation (e.g. incorporate in routine prenatal tests) rather than on specific target population
Comparing responses from different participants.	Duration for genetic counselling for participants from different specialty Waiting time for phlebotomy procedure Time spent and methods of conducting informed consent process

Key findings

Overall satisfaction levels were comparable to evaluation studies on newly established genetic services in foreign countries and local patient satisfaction surveys

Most of participants claimed advancing scientific research and helping others a big motivating factor (Might have been volunteer bias)

Reliance of altruism and benevolence for recruitment of participants might be challenging when project was implemented territory-wide

Strengths and limitations

First and only evaluation study on HKGP, provide much insights on roll-out of genomic projects
A mixed-methods approach allowed us investigation of project with rigorous breadth and depth

Due to constraint in time, no patient representatives/ patient groups were consulted in survey design process □ Would be included for subsequent phases

Majority of participants who had time for focus group interview were middle-aged/ elderly, they had hard time using zoom □ Would provide better instructions

NPS was widely used in business settings but rarely validated in healthcare studies □ Look for alternatives in future reporting of results, perhaps analysis of pre-post responses

1 Engage Stakeholders
2 Describe the program
3 Design evaluation methods
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5 Justify conclusions
6 Put to use and lessons learned

Way forward

Final phase of the evaluation cycle. Final report by Oct 2025

- **Evaluation of the multi-dimensional impact on clinical utility, psycho-social-economic outcomes, and patient empowerment**
  Assess the post-test clinical utility of WGS for the HKGP using the C-GUIDE and validate the C-GUIDE in Hong Kong via HKGP (HKCH)
- **Psycho-Socio-Economic outcomes on patients and family**
  Awareness/Attitudes, Health-related quality of life (EQ-5D), Psychosocial well-being, Willingness to pay/preferences, Healthcare and community support (QMH)
- **Patient empowerment through genetic counselling**
  Apply a well-recognised and validated tool, the 24-item Genetic Counselling Outcome Scale (GCOS-24), to evaluate patient empowerment (PWH)

Key takeaways

- Commissioned mixed-methods study (survey + focus group)
- 9/11 indicators achieved satisfaction level of 80% or above.
- Most participants stated the primary reason for engaging in the HKGP was to help **advance scientific research** and **benefit future patients** *rather than* expecting direct benefits to themselves or relatives.
- Participants cited **privacy, price, complexity, utility, sensitivity of results, and confidence in test providers** as primary reasons not engaging in alternative genetic tests elsewhere.
- Satisfaction levels were on par with foreign genome projects

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Experiences of Participants with Undiagnosed Diseases and Hereditary Cancers during the Initial Phase of the Hong Kong Genome Project: A Mixed-Methods Study

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Q&A