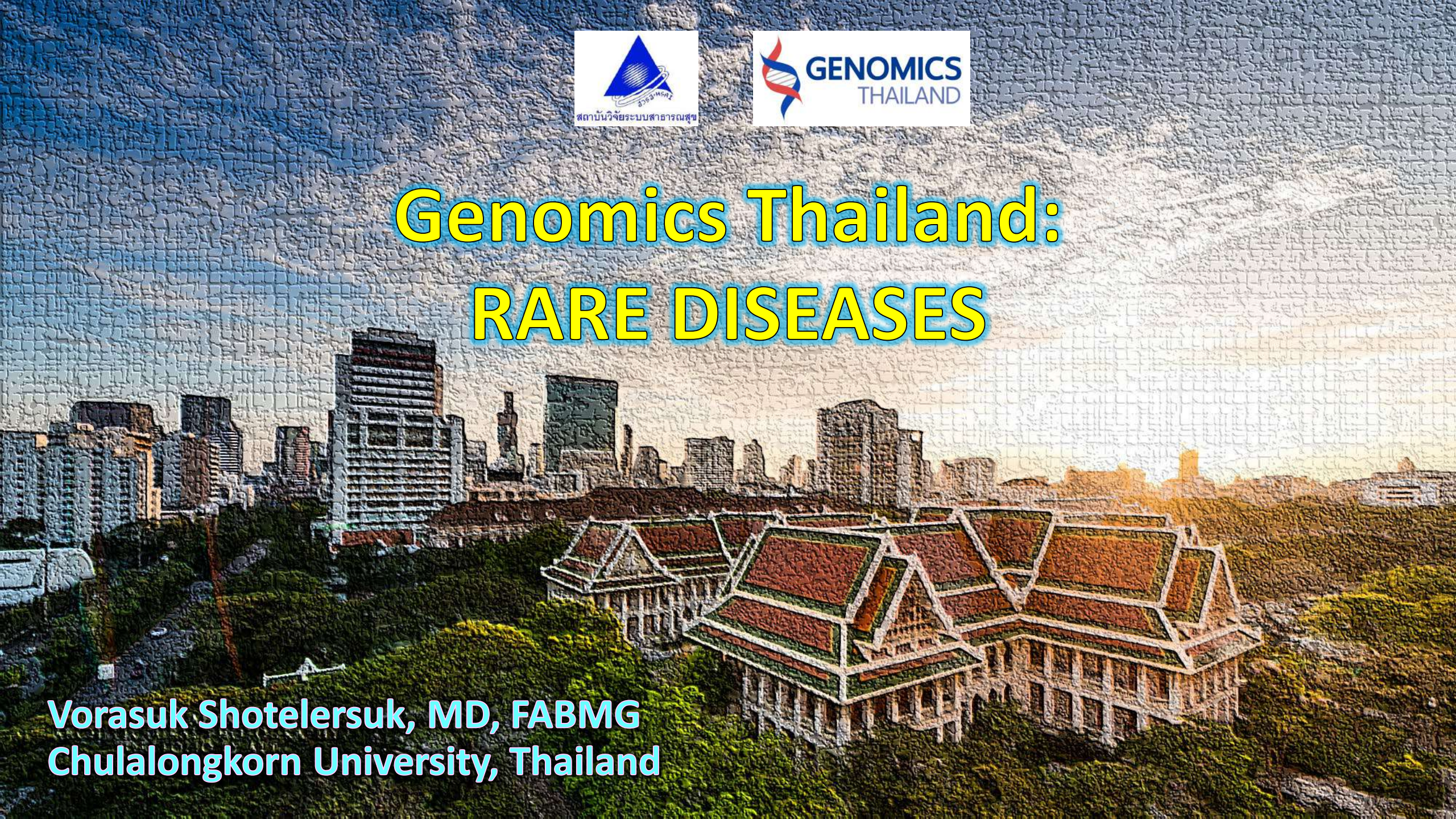

สถาบันวิจัยระบบสาธารณสุข
GENOMICS
THAILAND
Genomics Thailand:
RARE DISEASES
Vorasuk Shotelersuk, MD, FABMG
Chulalongkorn University, Thailand

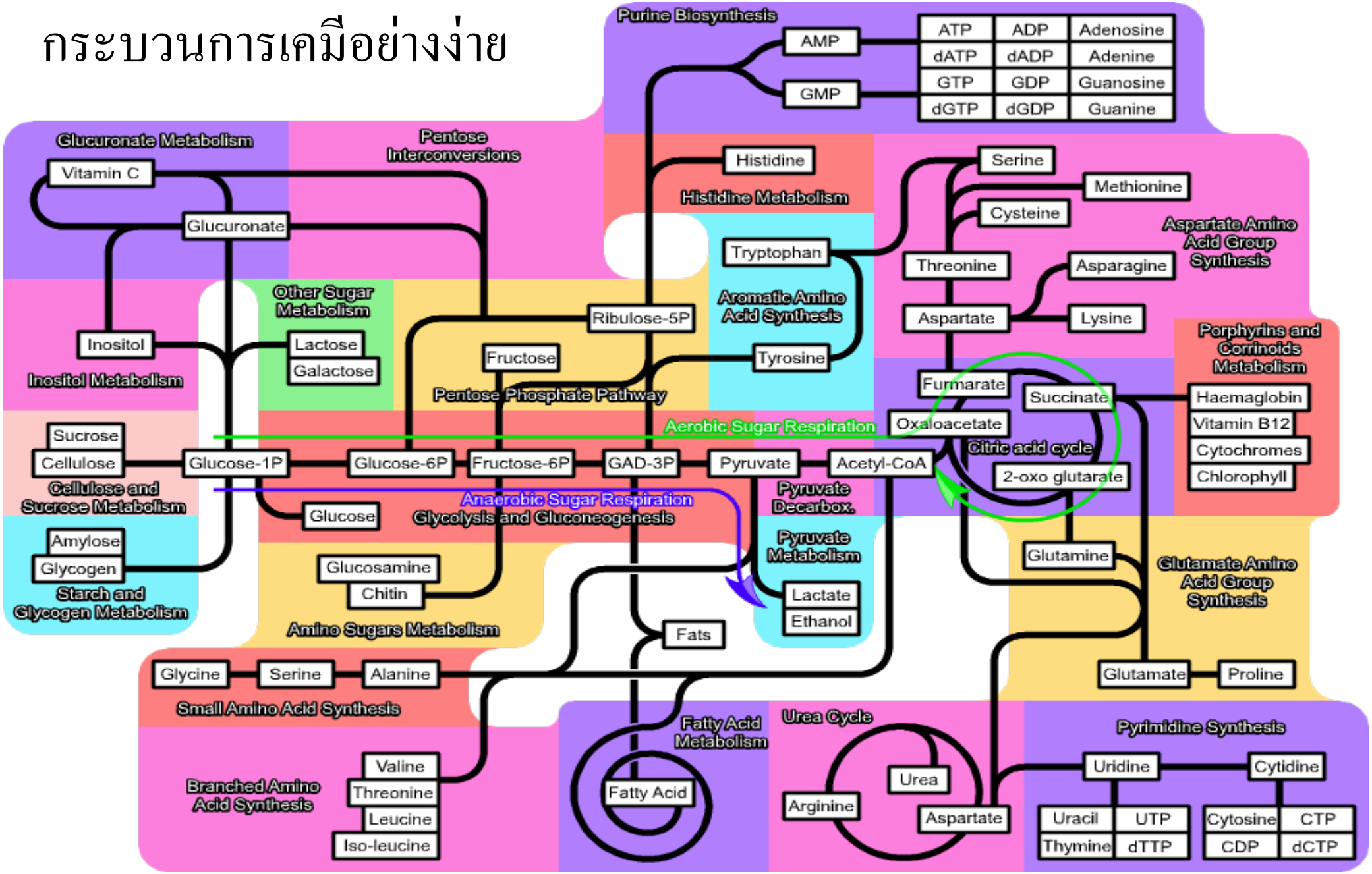
อาการ: TV เปิดไม่ติด

สาเหตุ

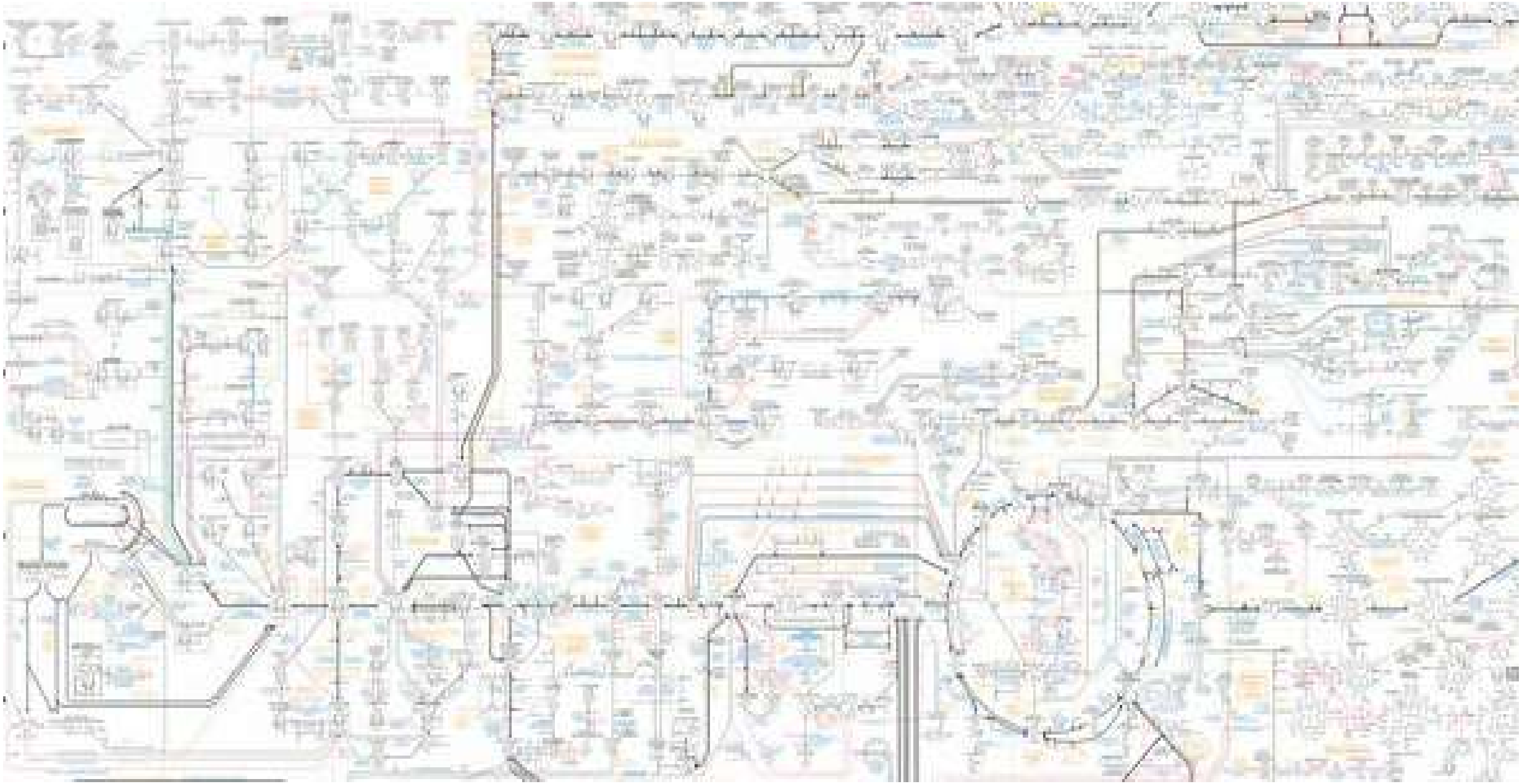
1. ไฟไม่เข้าบ้าน
2. รางปลั๊กเสีย
3. ไม่เปิดสวิทช์รางปลั๊ก
4. ฟิวส์รางปลั๊กขาด
5. Remote เสีย
6. ไม่ได้ใส่ถ่าน remote
7. ถ่าน remote เสื่อม
8. ไม่ได้เสียบปลั๊กทีวี
9. .....

เด็กพัฒนาการช้า

กระบวนการเคมีอย่างง่าย



กระบวนการเท่าที่มนุษย์เข้าใจใน 1 เซลล์



อาการ
100 อย่าง

พัฒนาการช้า

มองไม่เห็น

หูหนวก

ตัวเหลือง

ตัวซีด

เหนื่อยงาน

สาเหตุ
10,000 โรค

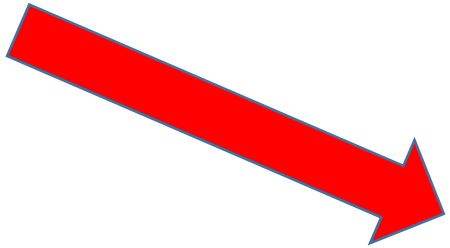
พัฒนาการช้า

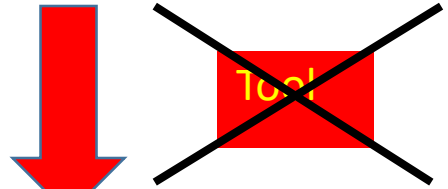
1,000 ยีน

แต่ละโรค-หายาก ทั้งกลุ่มโรค-พบบ่อย

< 1 in 2,000
> 7,000 distinct diseases

80% have
a genetic component





5-8% of the population
(Eurordis, 2005)
(5 M people in Thailand)

7 years to a diagnosis

แต่ละโรค-หายาก ทั้งกลุ่มโรค-พบบ่อย

< 1 in 2,000
> 7,000 distinct diseases

80% have
a genetic component

5-8% of the population
(Eurordis, 2005)
(5 M people in Thailand)

Days to a diagnosis

Benefits: patients, families, economy

World Health Organization

Science -> Public Health

WHO Science Council 2021

Accelerating access to genomic technologies for global health

The WHO Science Council functions to:

- Identify emerging issues on science and technology with the potential for direct or indirect impact on global health

Genomics Technologies

The Science Council has identified genomic technologies as having significant implications for public health, given their broad applications across health and disease states and throughout the human lifespan.

The NEW ENGLAND JOURNAL of MEDICINE

N ENGL J MED 385;20 NEJM.ORG NOVEMBER 11, 2021

ORIGINAL ARTICLE

100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care — Preliminary Report

CONCLUSIONS

Our pilot study of genome sequencing in a national health care system showed an increase in diagnostic yield across a range of rare diseases. (Funded by the National Institute for Health Research and others.)

Genomics Thailand

1. rare (difficult-to-diagnosed) diseases
2. Cancer
3. Pharmacogenomics
4. Non-communicable diseases
5. Infectious diseases





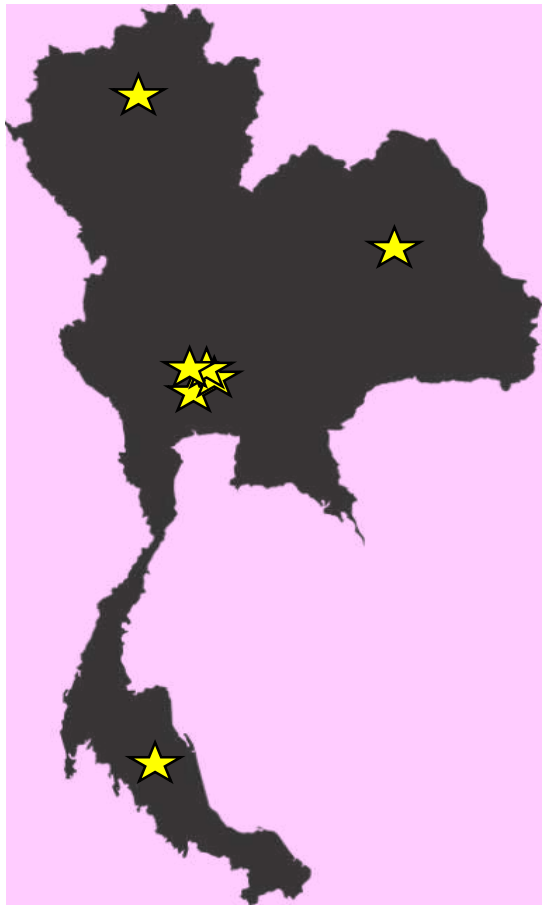
Thailand Rare & Undiagnosed Disease Network (T-RUN): 9 medical centers

Chulalongkorn:
Kanya, Thantrira

Children's:
Chulaluck

Phramongkutklao
Boonchai

Siriraj
Achara



ChiangMai
Pimlak,
Rekwan,
Maliwan

Rama:
Duangrurdee,

KhonKhan
Khunton

Thammasart
Kitiwan

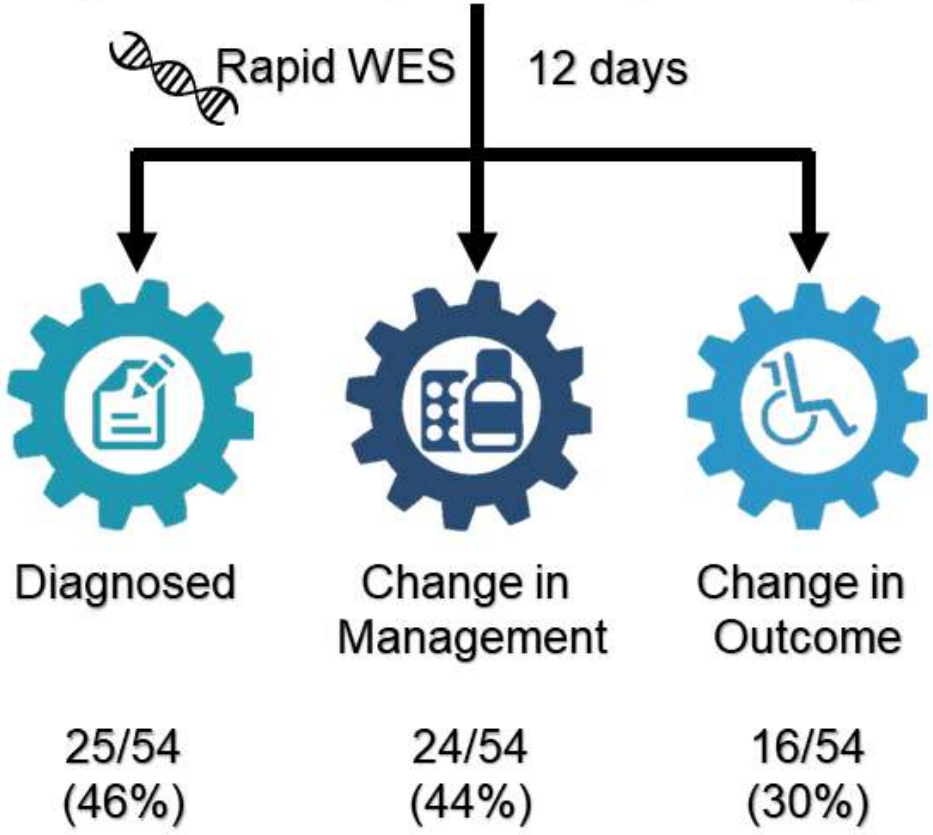
Songkla
Somjit

Leave No One Behind

ผู้ป่วยที่มีอาการรุนแรงและเฉียบพลัน

Exome sequencing as the first-tier investigation

[54 critically ill Thai patients]



SHORT REPORT

CLINICAL GENETICS WILEY

11 hospitals

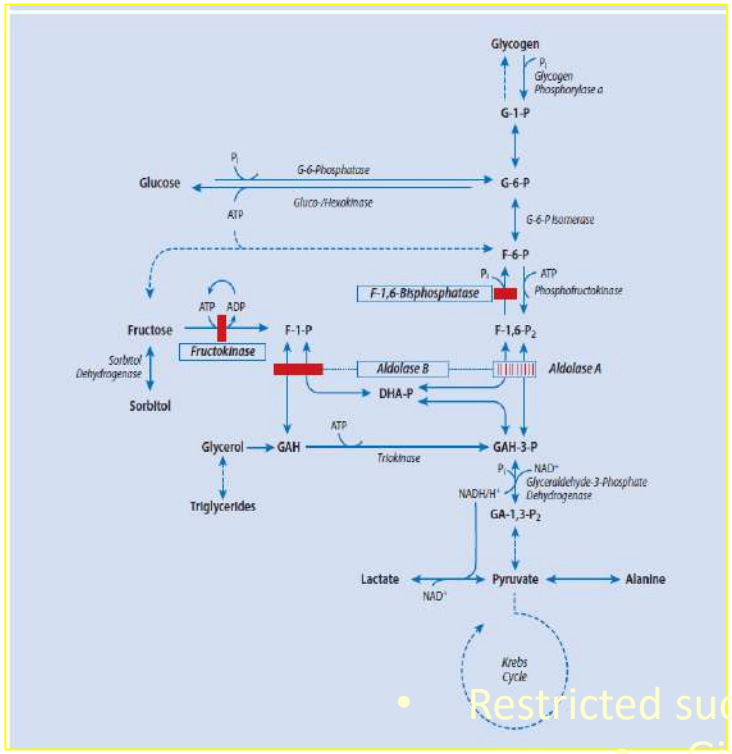
Clinical Genetics. 2021;100:100–105.

Rapid exome sequencing as the first-tier investigation for diagnosis of acutely and severely ill children and adults in Thailand

Wuttichart Kamolvisit[1,2] | Prasit Phowthongkum[2,3] | Ponghatai Boonsimma[1,2] | Chulaluck Kuptanon[4,5] | Kitiwan Rojnueangnit[6] | Duangrurdee Wattanasirichaigoon[7] | Mongkol Chanvanichtrakool[8] | Chutima Phuaksaman[9] | Pattara Wiromrat[10] | Chalurmpon Srichomthong[1,2] | Chupong Ittiwut[1,2] | Chureerat Phokaew[1,2] | Rungnapa Ittiwut[1,2] | Adjima Assawapitaksakul[1,2] | Wanna Chetruengchai[1,2] | Aayalida Buasong[1,2] | Kanya Suphapeetiporn[1,2] | Vorasuk Shotelersuk[1,2]

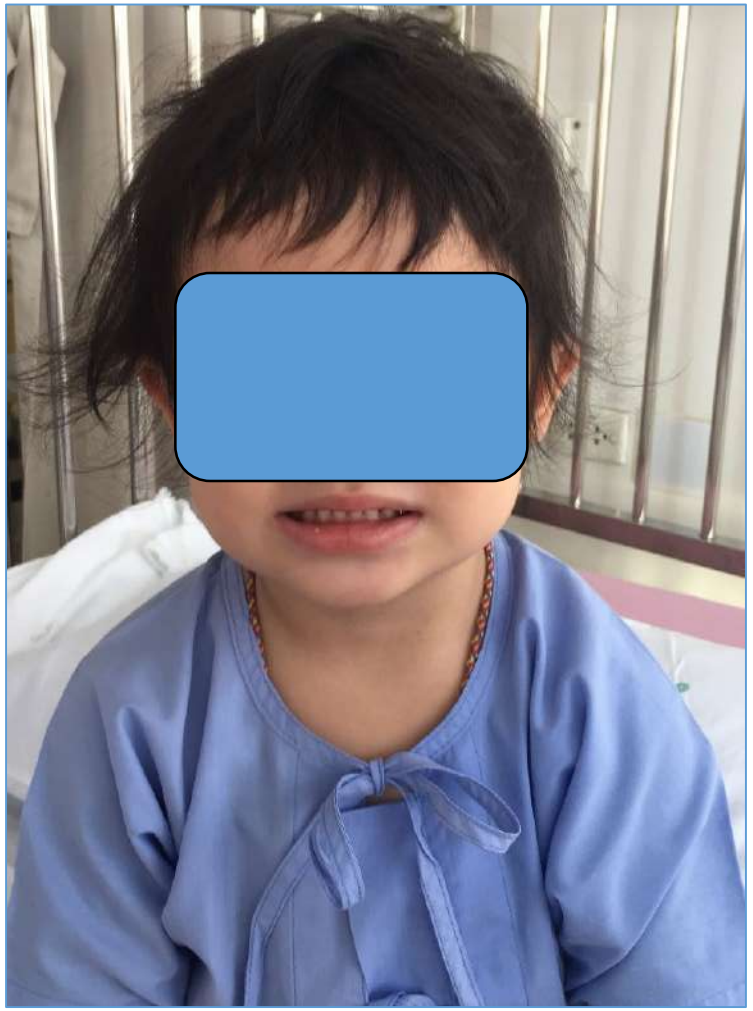
1st in LMIC, in adults, as a network

Fructose 1,6 Biphosphatase deficiency
(Incidence: 1:900,000)

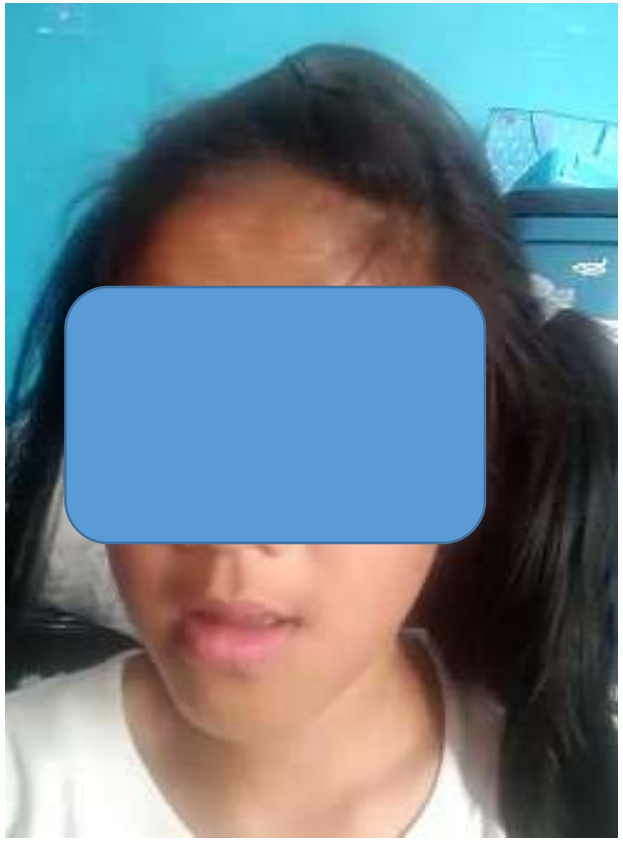


- Restricted sucrose, fructose (fruits)
- Give glucose

Another life is saved!!!

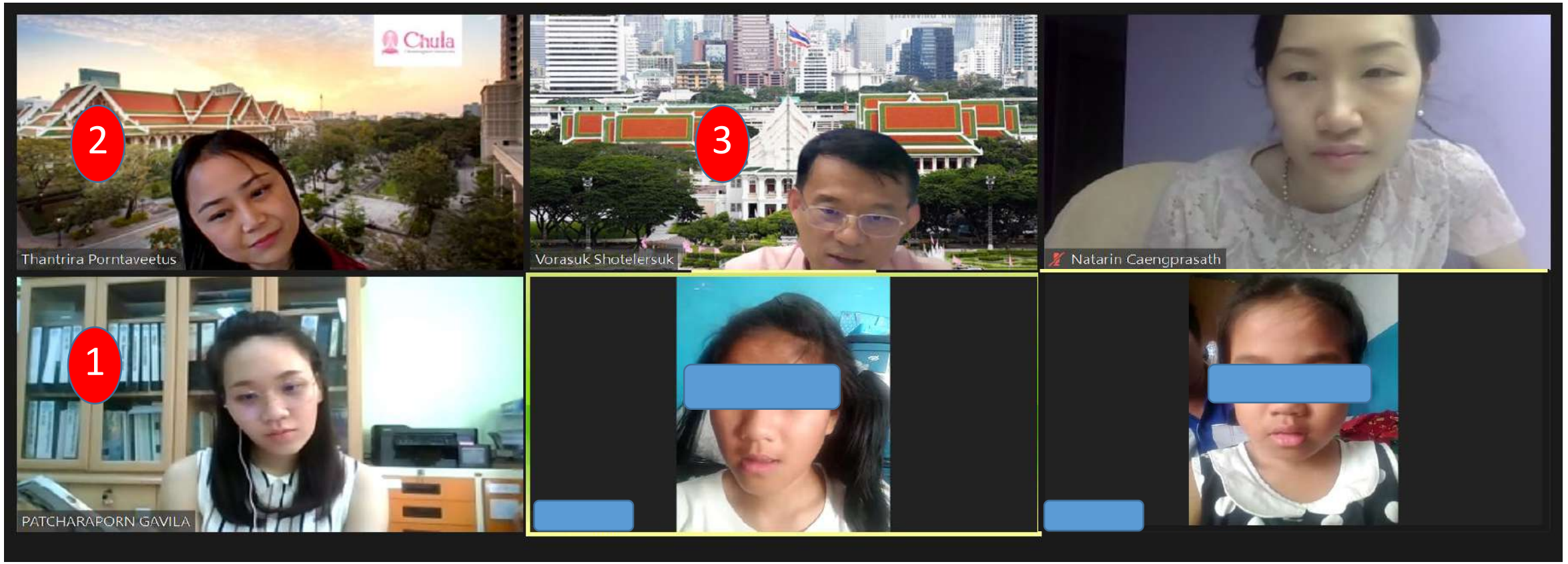


Two sisters from Chiang Mai



Amelogenesis imperfecta

Amelogenesis imperfecta + worsening eyesight



Time sensitive

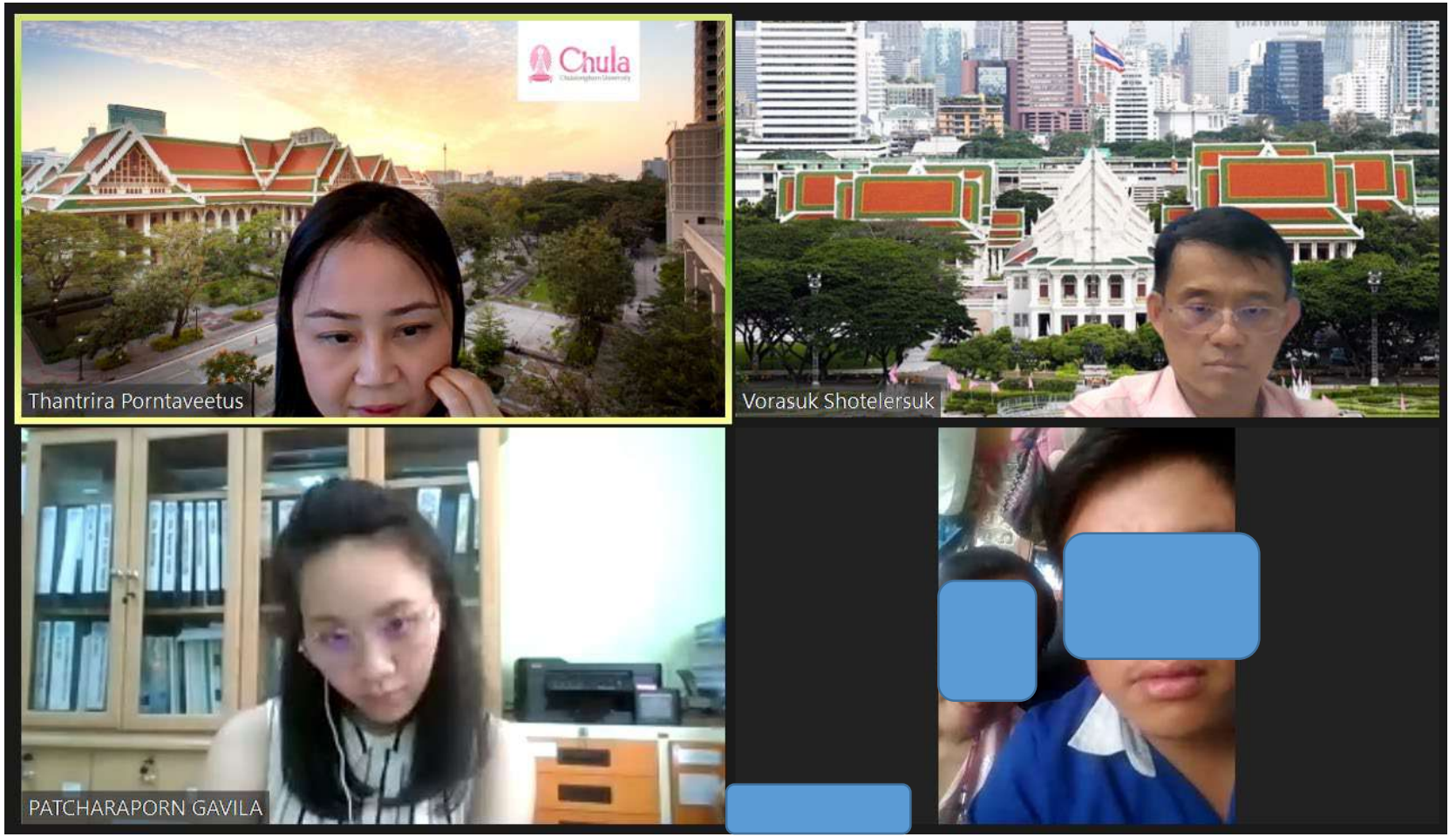
Early April 2021: Pregnant

Challenge: intended to terminate,

if the fetal disease status could not be determined

23rd April: 1st Zoom – offer molecular diagnosis in the sibs
(our 1st ever pre-test counselling via online)
(urgent: termination 23^{+6} week pregnant)

Parents agreed -> Exome sequencing

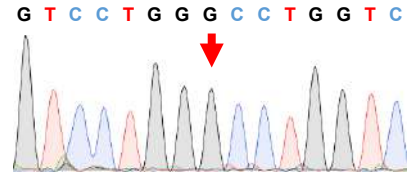


3rd May: 2nd zoom

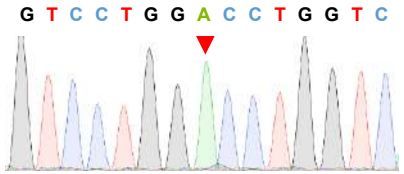
– delivered molecular diagnosis (our first ever)

- Prenatal diagnosis offered and contacted

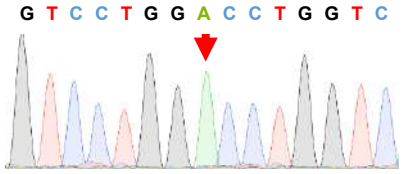
Jalili syndrome:
AI + cone and rod dystrophy



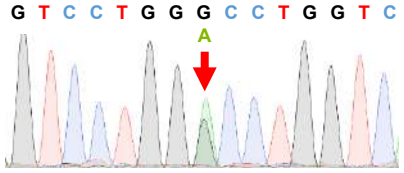
Control



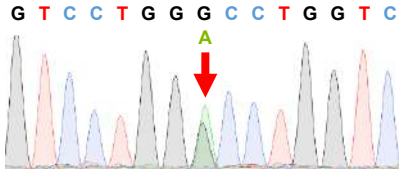
Patient 1



Patient 2



Father

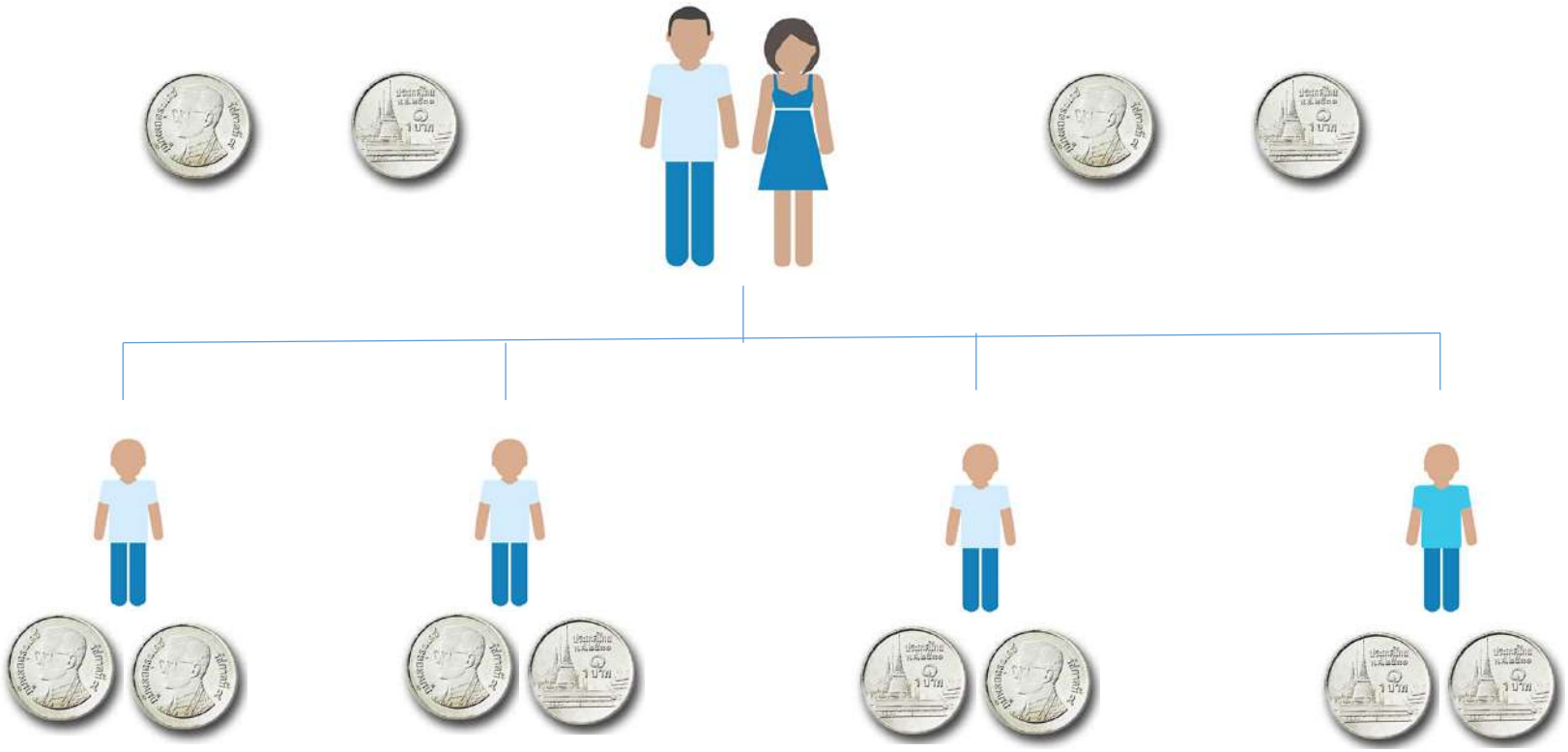


Mother

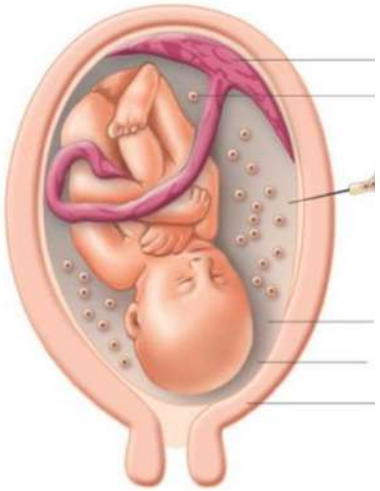
Sanger sequencing:

CNNM4 c.1475G>A

1st June: 3rd zoom
online pretest counselling for prenatal diagnosis (our first ever)



10th June 2021: Amniocentesis



23rd June: 4th zoom

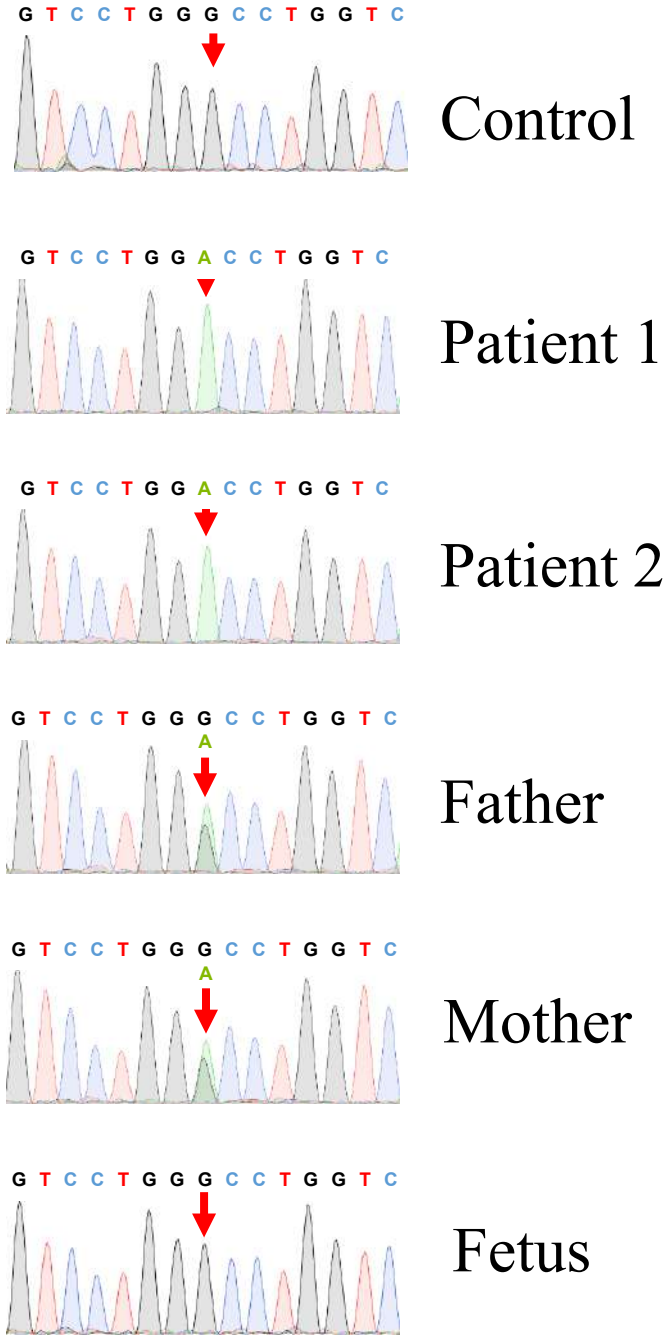
– delivered molecular diagnosis

- post-test counselling for PND (our 1st ever)

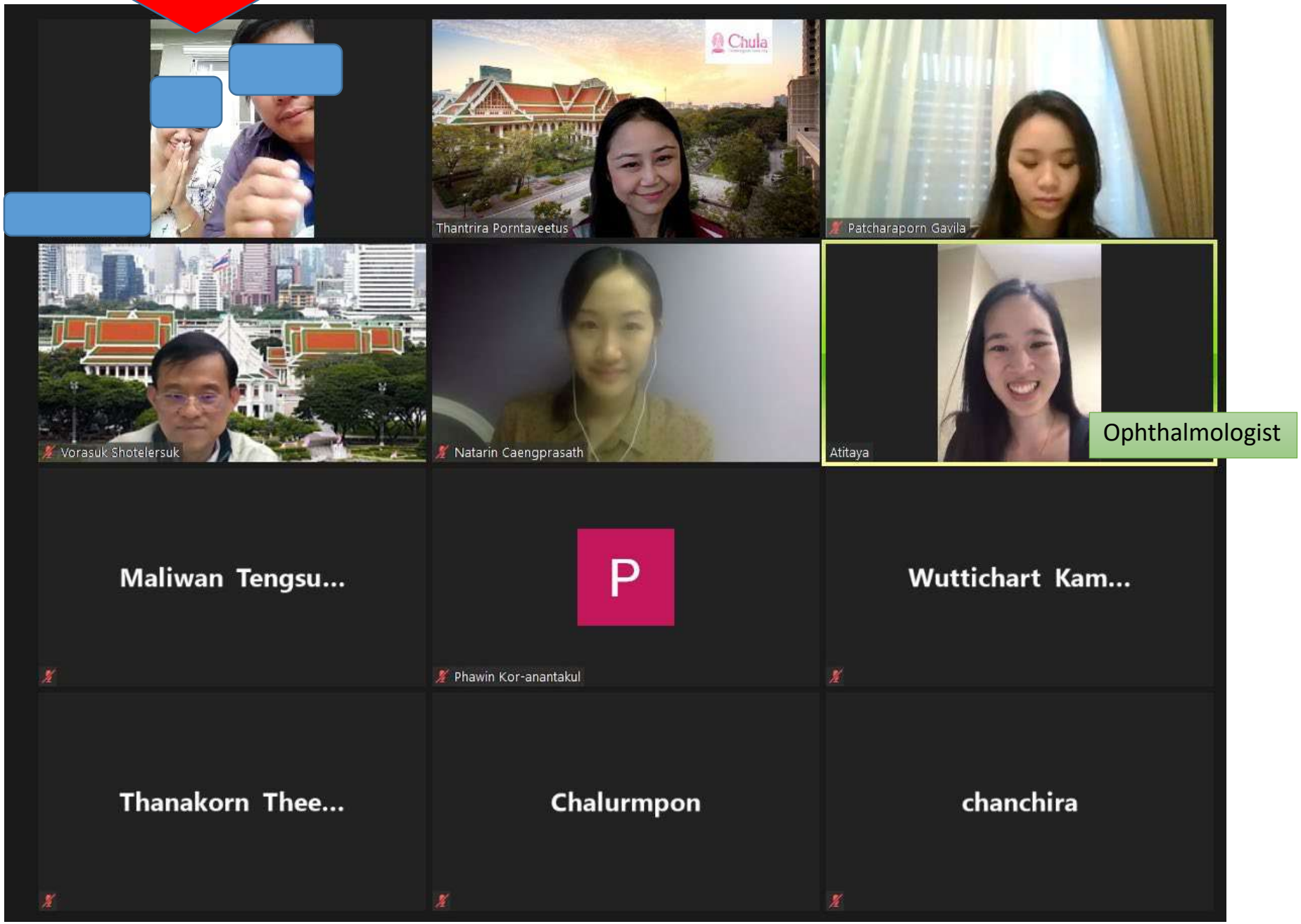
Very nervous

Homozygous: terminate

Otherwise: continue



รอยยิ้มและน้ำตาแห่งความอิ่มใจ สุขใจและภูมิใจ (EXOME Sequencing)



A network of Genomics Thailand saves another life.

สรุป

- โรคหายาก หมายถึง โรคที่พบ < 1 ใน 2,000 ประชากร
  แต่มีจำนวนโรคมาก รวมแล้ว เป็นกลุ่มโรคที่**พบบ่อย**
- Genomic Technology: เป็น**เครื่องมือใหม่ที่เหมาะ**กับ
  การวินิจฉัยโรคหายาก
- **Genomics Thailand** ทำให้ประเทศไทยสามารถนำศักยภาพของ
  Genomic technology มาใช้เพื่อ**วินิจฉัยโรคหายาก**
  ได้อย่างเต็มประสิทธิภาพ (-> save lives)
  (Genomics Thailand ระยะที่สอง **สุขภาพของคนไทย**)

Future

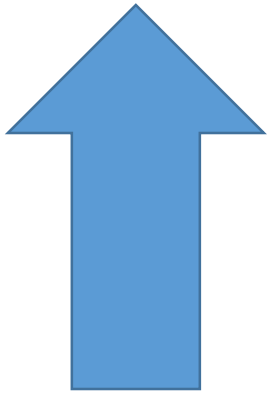
2 patients

3 healthy

Personalized Medicine

1

Read (+ Understand)

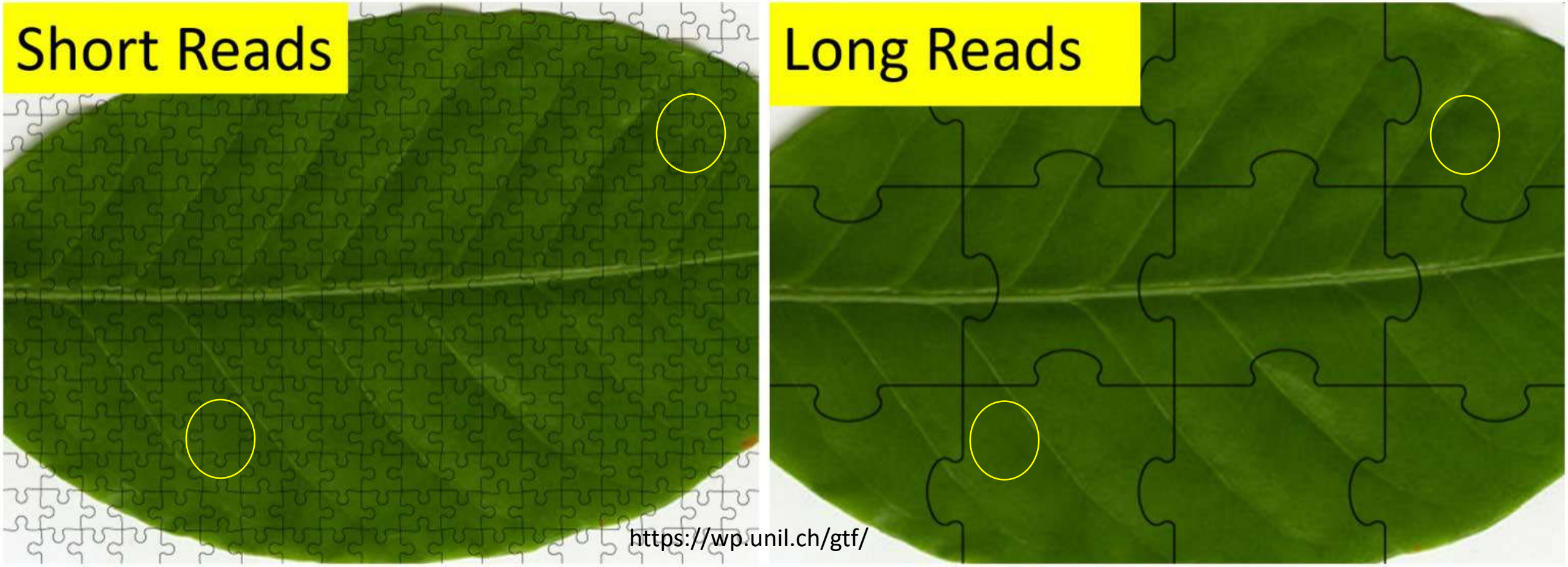


Write (+ Target)

Population-Average Medicine

Read | Understand | Write | Target

Tools | Applications



92% complete
Before 2021

HUMAN GENOME

100% complete
Nurk, BioRxiv May 2021

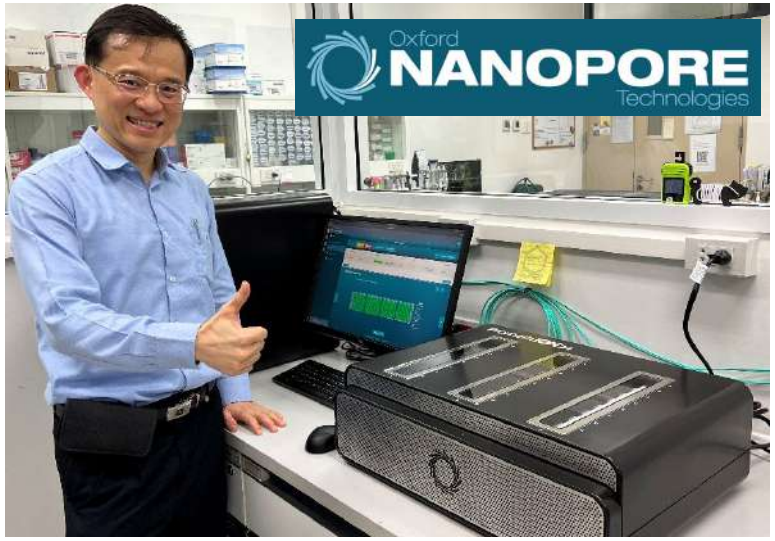
Short-read

Long-read

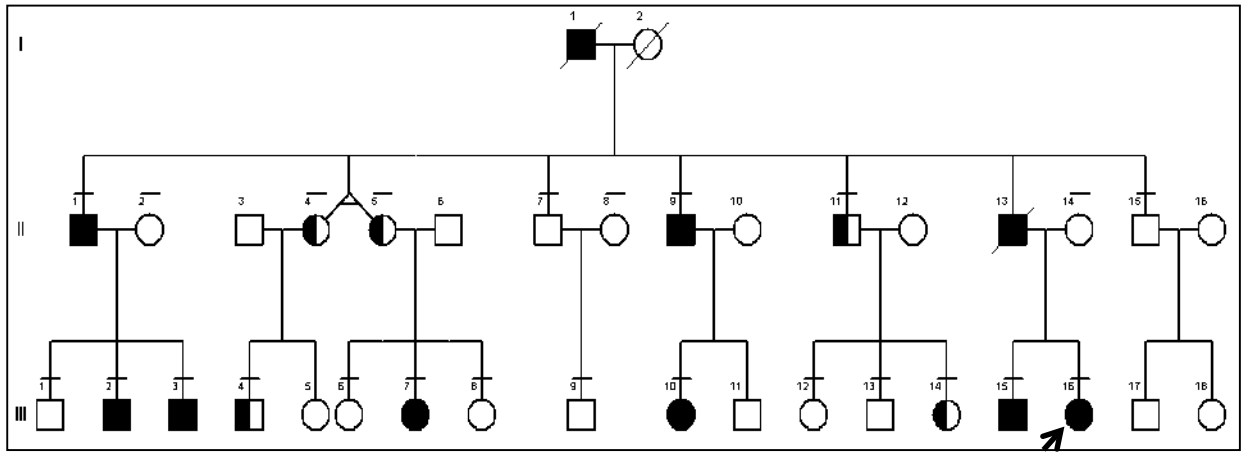
Linked-read
Single-cell

10x GENOMICS

Ultralong-read



2008 - 2019



doi:10.1093/brain/awz267 BRAIN 2019: 0; 1–7 | 1

BRAIN
A JOURNAL OF NEUROLOGY

IF 13.501

REPORT

TTTCA repeat insertions in an intron of *YEATS2* in benign adult familial myoclonic epilepsy type 4

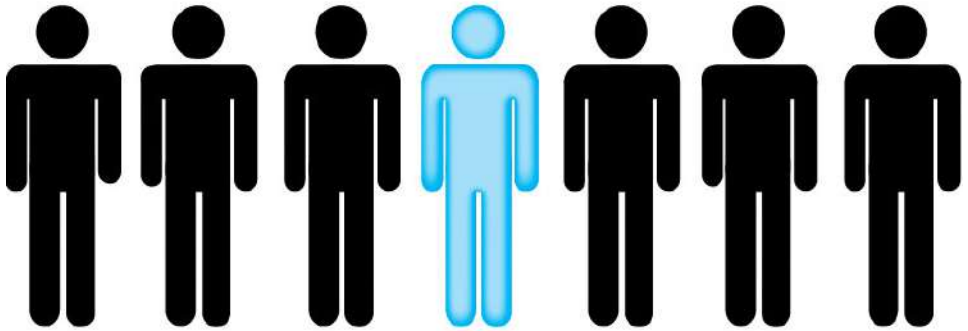
Patra Yeetong,[1] Monnat Pongpanich,[2,3] Chalurmpon Srichomthong,[4,5] Adjima Assawapitaksakul,[4,5] Varote Shotelersuk,[4,5] Nithiphut Tantirukdham,[1] Chaipat Chunharas,[6] Kanya Suphapeetiporn[4,5] and Vorasuk Shotelersuk[4,5]

20(TTTTA)

500(TTTTA)-500(TTTCA)

2

patients



Rare disease

15,000 -> .....

3

healthy

Couple sequencing

Newborn sequencing

Healthy adult sequencing

Thank you

Ultimate Goals

"maximize potential of the genomics technologies"
medical hubs
a leader in precision medicine

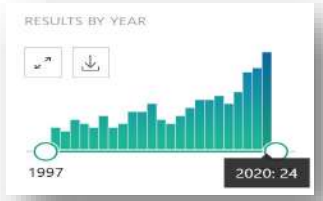
national and regional levels

social + economic impacts

Upscale

institution level

academic



1 publication/2 wk