Asia Pacific Society of Human Genetics

APSHG Newsletter

Volume 02 | Issue 01 | June 2021

Message from the President

Dear members, friends and colleagues of the Asia Pacific Society of Human Genetics, (APSHG),

Most of us will always remember where we were at the beginning of 2020 when governments of most countries in the world issued an unfamiliar message to their people to stay home, to work from home or that the city was under lockdown. This led to changes in our daily activities including the need for additional personal protective equipment such as wearing of face masks, frequent hand cleansing etc.

I am writing this report in the midst of the Covid-19 Pandemic and its anticipated second or third surge in countries including those in the Asia-Pacific region. I believe that the more challenged we are the more we learn from this situation. This is the logic of the brave and the keen observer. In fact, more than 91,132 articles on COVID-19 were published in 2020 with most having the highest number of citations on Google Scholar (PubMed). The Coronavirus 19 outbreak has increased research opportunities in the field of science, medicine and public health. Sequencing of the virus' genome has led to the discovery of more variants in different parts of the world resulting to an encouraging development of the most appropriate tools to combat the SARS-CoV-2 outbreak by epidemiologists, scientists, physicians, healthcare personnel including drug and vaccine companies.

I am pleased to note the increased membership of young scientists and geneticists in our society since 2019. This means a lot to us. It is our duty to help them as best as we can. An example would be the APSHG website which is used as a media for academic meetings and conferences on genetics, genomics, and genetic counselling.

In managing the society, the regular yearly executive meetings with the board members became quarterly online meetings since the beginning of 2020. Exchanges of ideas and collaborations have been well-received by members as evidenced by the success of the 3rd APSHG Summer School Meeting held virtually on August 2020 in Taiwan, and the publication of this newsletter. Also included in our 2021 activity calendar are the 14th APCHG Conference to be held in Indonesia in 2022 and the 4th Summer School to be held in Thailand late in 2021 or early 2022 either virtually or possibly using a hybrid platform.

I wish you all the best in this global hardship and hope to see you in the near future either virtually or in person in Indonesia and in Thailand.

Sincerely yours,

Than Sura

Thanyachai Sura MD MRCP
Professor of Medical Genetics
President of Asia Pacific Society of Human Genetics(APSHG)
President of Medical Genetics and Genomic Association (Thailand)

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In this issue, we focused on the Covid-19 pandemic that has led to a tragic loss of human lives worldwide and presents as an unprecedented challenge to public health, education, food safety and global connectivity. This pandemic has greatly impacted the global community as well as groups in society, including doctors, scientists, healthcare workers, other professions and the general public. Our organization was also not spared from the impact of this pandemic. To survive, we had to adapt and comply with new norms such as the way we deal with our work and the way we conduct our day-to-day tasks.

In adapting to the new norms, conferences, meetings and seminars are now conducted virtually to great success. In fact, many societies now prefer to organise virtual conferences as speakers and participants find it more convenient to join the discussions from the comfort of their office or home. This includes our society's next proposed virtual conference, the 14th Asia-Pacific Conference on Human Genetics (APCHG) though a firm decision has yet to be reached.

I would also like to highlight the World DNA Day, which is celebrated annually on 25th April to honour the completion of the Human Genome Project (HGP) in 2003, and the identification of the DNA double helix in 1953. After the United States Congress passed simultaneous resolutions designating April 25th as DNA Day, the World Human Genome Research Institute (NHGRI) included the celebration of DNA Day in its annual activity calendar.

The knowledge of DNA in humans has helped people to understand a variety of fundamental questions, including the total number of genes that we have, how our cells function, how diseases developed and what actually happens when we become sick. It is now possible to use a patient genetic profile to take action about disease control, his treatment plan, and medication — i.e. personalised medicine or precision medicine as it is popularly known. As more DNA data are being profiled and understood, precision medicine may soon become routine and a part of mainstream medicine. This is what we, as geneticists, dream of and look forward to.

Finally, we welcome any submission of news, announcements, opinions, or any information that may be of interest in your field of expertise.

Email your submission to me at zilfalil@gmail.com or to any member of the editorial board.

Prof. Zilfalil Bin Alwi
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APSHG Newsletter

Published by
Asia-Pacific Society of Human Genetics (APSHG)
| Secretariat Office: 100 Bukit Timah Road, KK Women's and Children's Hospital, Singapore 229899
| Email: apshg@apshg.info | Website: https://www.apshg.info/index.html

Covid-19 Pandemic in Malaysia

Malaysia is, at the moment, on its third wave of the COVID-19 pandemic (as of 31st May 2021). A total of 572,357 COVID-19 cases have been reported since the pandemic started early last year.

According to the Ministry of Health of Malaysia, the first wave of the pandemic started on January 25, 2020 and lasted for three weeks with only 22 reported cases. All these 22 patients recovered well. The first reported cases were three Chinese tourists who had close contact with an infected individual in Singapore before they entered Malaysia. Majority of cases recorded in the first wave were from China. Only two cases were discovered to be the outcome of local transmissions.

The second wave lasted for 4 months. It started on the 27th of February 2020 and ended on the 30th of June 2020. The Seri Petaling cluster was the largest which included a religious assembly at a mosque in Seri Petaling, Kuala Lumpur. The handling of these two waves by the Ministry of Health Malaysia was excellent. It was reported that there were 10,145 cases with 9,293 cases recovered during the second wave. To control the spread of the disease, the Movement Control Order (MCO) was implemented during the second wave due to the sudden increase in number of cases.

A special committee for Ensuring Access to COVID-19 Vaccine Supply (JKJAV) was established and co-chaired by the Minister of Health and the Minister of Science, Technology, and Innovation to ensure the country's accessibility to COVID-19 vaccine supplies. JKJAV also acts as the major committee in the preparation, implementation, and supervision of the entire National COVID-19 Immunization Programme. The National COVID-19 Immunization Programme details the national plans, vaccine supply sourcing plans, delivery initiatives, and monitoring to combat the COVID-19 pandemic.

Malaysia managed to secure 66.7 million doses of COVID-19 vaccines as of February 2021 via the COVAX Facility and advance orders from five vaccine manufacturers namely Pfizer-BioNTech, AstraZeneca, Sinovac, CanSino Biologics, and Sputnik V. The Pfizer-BioNTech vaccine received conditional permission from the Malaysian Drug Control Authority (DCA) and the Malaysian National Pharmaceutical Regulatory Agency (NPRA) on January 8, 2021. The remaining four COVID-19 vaccine candidates also sought NPRA acceptance. According to the NPRA, Malaysia will acquire vaccines from these five suppliers in stages beginning February 2021.

Since the 26th of February 2021, about 126,000 people in the country were vaccinated daily with the COVID-19 vaccine through the COVID-19 National Immunization Plan. The government aims to vaccinate 80% of the population or an equivalent of 26 million individuals.

As part of an ongoing effort to fight Covid-19, the Malaysian government, through the Ministry of Science, Technology and Innovation (MOSTI), allocated a grant of RM70,000 to the Malaysia Genome Institute (MGI) to conduct full genome sequencing and bioinformatics analysis to detect mutations in the genome from coronavirus samples (Covid-19). MGI has partnered with the Public Health Laboratory, National Institute of Health and Medical Research Institute of the Ministry of Health Malaysia to perform genome-wide sequencing of Malaysian COVID-19 samples to develop complete ribonucleic acid genome sequence data which can be matched for correlation with the COVID-19 strain detected within and outside the country. There were 416 genomes from Malaysia that have been registered in the middle of March 2021 at the Global Initiative for Sharing All Influenza Data (GISID) which included 44 genomes from MGI. MGI reported that notable mutations among Malaysians were detected namely D614G, VUI 202012/01, and G/484K.V3 with 69%, 0.48% and 0.48% frequency, respectively.

The COVID-19 Research Group from the Universiti Malaya (UM) is another group that sequenced four SARS coronavirus 2 (SARS-CoV-2) genomes from Malaysia during the second wave of the pandemic and discovered unusual mutations that indicated local evolution of the virus. They conducted full genome sequencing of SARS-CoV-2 strains obtained directly from nasopharyngeal swabs of four patients in Kuala Lumpur, Malaysia. Their results provided detailed comprehension of the SARS-CoV-2 molecular epidemiology in Malaysia.[1]

Another research group from the Universiti Malaysia Pahang (UMP) in collaboration with the International Islamic University Malaysia (IIUM) also conducted a research on the SARS-CoV-2 genome. On April 2, 2020, an asymptomatic patient provided nasopharyngeal and oropharyngeal swabs as well as a sputum extract. SARS-CoV-2 was detected using real-time reverse transcriptase (RT)-PCR. The genome sequence was stored in public databases such as the National Center for Biotechnology Information (NCBI) and GISID.[2]

The Covid-19 pandemic in Malaysia is not yet over. We are still fighting this pandemic. The government has started taking more strict control measures in order to prevent further outbreak. With everyone's support including support from the Ministry of Health Malaysia, frontline workers, healthcare professionals, and the public community, there is hope for us to win the battle and to break the Covid-19 chain in the country.

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Covid-19 Pandemic in Thailand

On 13 January 2020, Thailand was the first country to report a Covid-19 case outside China. The country reported a cumulative total of 128,116 confirmed cases and 759 deaths from the disease as of 23 April 2021.

Thailand was relatively successful in containing the pandemic throughout most of 2020 but is currently experiencing an uncontrolled resurgence since April 2021. The initial wave of infections, mostly traced to nightlife venues and a boxing match in cities such as Bangkok, peaked on 22 March 2020 at 188 newly confirmed cases per day. The outbreak subsided by May when preventive measures were implemented. The country reported almost no locally transmitted infections until December 2020.

A second surge of infections primarily clustered around large migrant worker communities in the Samut Sakhon Province. This new outbreak spread to many provinces peaking at 959 cases reported on 26 January 2021 before partially subsiding in February 2021.

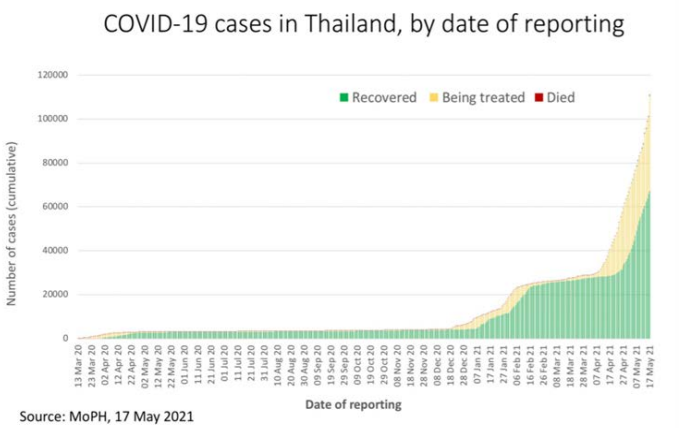
However, in April 2021, a new wave of infections originating from Bangkok's nightlife venues rapidly spread in the city and throughout the country. It was identified to be secondary to the highly transmissible B.1.1.7 variant first reported in the United Kingdom. By mid-April, over a thousand cases were identified daily thereby causing a shortage of hospital beds as government policy required admission of all confirmed cases. The situation worsened by the end of April after the Songkran holidays. The Songkran Festival takes place in April every year from the 13th to the 15th with many Thai people making their way to their hometowns to spend time with their families. It is Thailand's most famous festival marking the beginning of the traditional Thai New Year. Thailand is undoubtedly presently experiencing its biggest and most challenging third wave of Covid-19 infections.

The Thai government and the Centre for COVID-19 Situation Administration (CCSA) which was established during the first outbreak in March 2020, work in conjunction with the Department of Disease Control by tracing and actively finding new cases in most worker communities in Bangkok, the perimeter areas, and in the main provinces of Thailand's border areas. Restricted activity measures have been implemented in varying degrees throughout the country, with public venues and businesses ordered to close. All commercial international flights were suspended, especially those from India and Africa. In spite of the state of emergency still in effect, the public has cooperated relatively well with these health advisories. The country's robust public health infrastructure is also being credited as a major contributing factor to the country's relatively successful initial response.[4,5]

The importance of vaccination in the control of the pandemic has been repeatedly emphasized and implemented by the Thai government. Vaccinations began at the end of February 2021, mostly limited to healthcare workers and mainly using the CoronaVac vaccine imported from China's Sinovac Biotech and the Astra Zeneca vaccine from Europe. Majority of the country's vaccine supply relies heavily on the Oxford–AstraZeneca vaccine through a manufacturing deal secured by the Thailand Siam Bioscience company. First batches are expected to arrive in June 2021. At the moment, 2.8 million Thai individuals have been vaccinated of whom 2.1 million are fully vaccinated having received their second dose in a 2-dose vaccine series. By early June, all 6 million doses of AstraZeneca vaccines are supposed to be administered to the Thai population and subsequently by the end of 2021, 70% equivalent or 49 million of the entire Thai population would have been vaccinated.

The Thai government has set the National COVID-19 Vaccination plan as one of its priorities to combat this outbreak. It has allocated 900,000,000 Baht (USD 30 million) for the Ministry of Science and Technology in conjunction with the country's top universities to conduct the Coronavirus 19 genomic study and to develop vaccines. There are two vaccine development programs both of which are currently in their phase 3 studies (Rakrungtham K. and Co.).

The COVID-19 pandemic has affected every nation in the world and it is not yet over. There have been 167,070,797 Corona virus cases and 3,469,436 deaths worldwide as of 23 May 2021. It is our sincere hope that with the support of everyone (the CCSA, the government sector, and the Thai people), we can beat this pandemic and return back to our lives.



Source: MoPH, 17 May 2021

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Covid-19 Pandemic in the Philippines

Since the start of the pandemic until June 6, 2021, the Philippines has had 1,262,273 confirmed COVID-19 cases with 21,732 deaths.[1] Vaccination is a slow undertaking given the lack of resources and logistical issues to deliver vaccines to 7,640 islands for approximately 110 million people. Despite these barriers, a total of 4,875,342 vaccine doses have been administered as of May 28, 2021.[1]

The national government implemented a COVID-19 Vaccination Program Prioritization Framework and vaccine rollout commenced in the first quarter of 2021. It initially targeted the frontline healthcare workers, senior citizens, and people with existing health conditions. In an effort to curtail COVID-19 transmission and jumpstart the economy, the country now intends to vaccinate the 35 million Filipino workforce. So far, the country has received 9 million COVID-19 vaccines with around 22 million additional COVID-19 doses expected to arrive in the next 2 months. More needs to be done, however, before it can meet its goal of immunizing 70 million people before the year ends.[2]

The Philippine government has established a multi-sectoral response through the Interagency Task Force (IATF) on Emerging Infectious Diseases chaired by the Department of Health (DOH) which keeps the public updated with the government's strategic plans to keep everyone healthy and safe. The government has since enforced a community quarantine in Metro Manila and in other parts of the country, expanded its testing capacity, and bolstered the country's healthcare system.

As of May 29, 2021, a total of 7,547 COVID-19-positive samples have undergone sequencing by the University of the Philippines-Philippine Genome Center (UP-PGC) and the University of the Philippines-National Institutes of Health (UP-NIH). The Philippines has confirmed 2,494 total cases with variants monitored by the DOH. There are, at present, only 26 active cases of the more infectious COVID-19 variants that were first detected in the United Kingdom (B117), South Africa (B1351), and in the Philippines (P3).[2]

Meantime, the impact of COVID-19 to the physical and mental health of the Filipino people is devastating. With strict community quarantine, income source and children's education have been seriously compromised. To also highlight, the Filipino culture honors living in a multigenerational household. As such, the virus easily spreads to several family members in a matter of days which then depletes the family's financial resources along with prolonged chronic stress of worrying for everyone's health.

The country clearly continues to face many difficulties. An intensified effort to prevent, control transmission of infections through proper surveillance efforts, rigorous contact tracing, isolation of cases, quarantine of contacts while ensuring timely and adequate laboratory and therapeutic access continue to be the primary public health concern.

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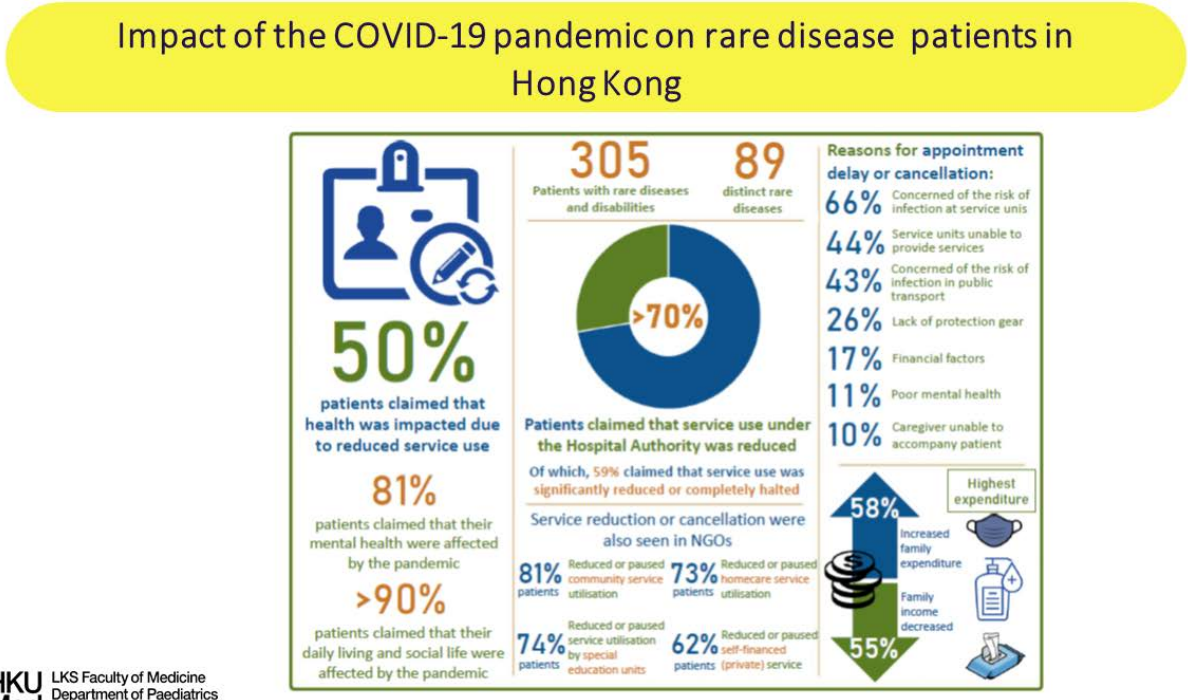
Covid-19 Pandemic in Hong Kong



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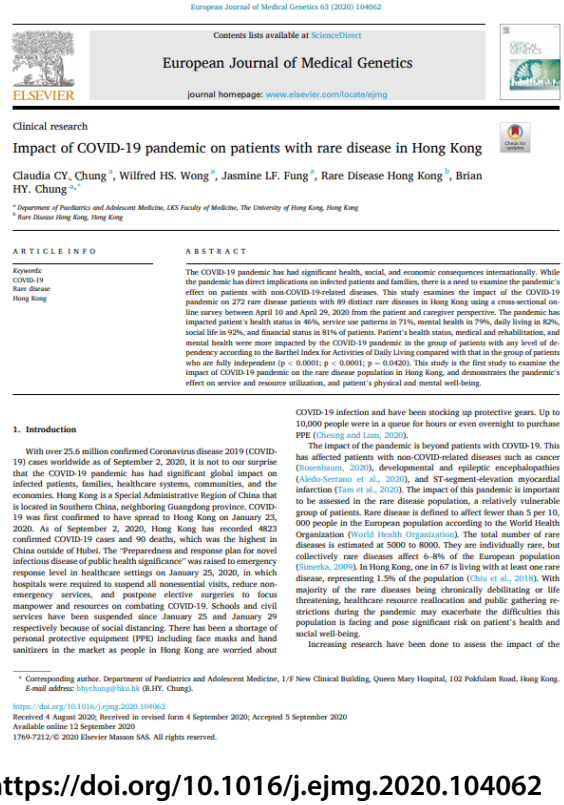
Accepted in *Eur J Med Genet* 2020

Abstract

The COVID-19 pandemic has had significant health, social, and economic consequences internationally. While the pandemic has direct implications on infected patients and families, there is a need to examine the pandemic's effect on patients with non-COVID-19-related diseases. This study examines the impact of the COVID-19 pandemic on 272 rare disease patients with 89 distinct rare diseases in Hong Kong using a cross-sectional online survey between April 10 and April 29, 2020 from the patient and caregiver perspective. The pandemic has impacted patient's health status in 46%, service use patterns in 71%, mental health in 79%, daily living in 82%, social life in 92%, and financial status in 81% of patients. Patient's health status, medical and rehabilitation, and mental health were more impacted by the COVID-19 pandemic in the group of patients with any level of dependency according to the Barthel Index for Activities of Daily Living compared with that in the group of patients who are fully independent ($p < 0.0001$; $p < 0.0001$; $p = 0.0420$). This study is the first study to examine the impact of COVID-19 pandemic on the rare disease population in Hong Kong, and demonstrates the pandemic's effect on service and resource utilization, and patient's physical and mental well-being.



European Journal of Medical Genetics 63 (2020) 104062

Contents lists available at ScienceDirect

European Journal of Medical Genetics

journal homepage: www.elsevier.com/locate/ejmg

Clinical research

Impact of COVID-19 pandemic on patients with rare disease in Hong Kong

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ARTICLE INFO

Keywords:
COVID-19
Rare disease
Hong Kong

ABSTRACT

The COVID-19 pandemic has had significant health, social, and economic consequences internationally. While the pandemic has direct implications on infected patients and families, there is a need to examine the pandemic's effect on patients with non-COVID-19-related diseases. This study examines the impact of the COVID-19 pandemic on 272 rare disease patients with 89 distinct rare diseases in Hong Kong using a cross-sectional online survey between April 10 and April 29, 2020 from the patient and caregiver perspective. The pandemic has impacted patient's health status in 46%, service use patterns in 71%, mental health in 79%, daily living in 82%, social life in 92%, and financial status in 81% of patients. Patient's health status, medical and rehabilitation, and mental health were more impacted by the COVID-19 pandemic in the group of patients with any level of dependency according to the Barthel Index for Activities of Daily Living compared with that in the group of patients who are fully independent ($p < 0.0001$; $p < 0.0001$; $p = 0.0420$). This study is the first study to examine the impact of COVID-19 pandemic on the rare disease population in Hong Kong, and demonstrates the pandemic's effect on service and resource utilization, and patient's physical and mental well-being.

1. Introduction

With over 25.6 million confirmed Coronavirus disease 2019 (COVID-19) cases worldwide as of September 2, 2020, it is not to our surprise that the COVID-19 pandemic has had significant global impact on infected patients, families, healthcare systems, communities, and the economies. Hong Kong is a Special Administrative Region of China that is located in Southern China, neighboring Guangdong province. COVID-19 was first confirmed to have spread to Hong Kong on January 23, 2020. As of September 2, 2020, Hong Kong has recorded 4823 confirmed COVID-19 cases and 90 deaths, which was the highest in China outside of Hubei. The "Preparedness and response plan for novel infectious disease of public health significance" was raised to emergency response level in healthcare settings on January 25, 2020, in which hospitals were required to suspend all nonessential visits, reduce non-emergency services, and postpone elective surgeries to focus manpower and resources on combating COVID-19. Schools and civil services have been suspended since January 25 and January 29 respectively because of social distancing. There has been a shortage of personal protective equipment (PPE) including face masks and hand sanitizers in the market as people in Hong Kong are worried about COVID-19 infection and have been stocking up protective gears. Up to 10,000 people were in a queue for hours or even overnight to purchase PPE (Cheung and Lum, 2020).

The impact of the pandemic is beyond patients with COVID-19. This has affected patients with non-COVID-related diseases such as cancer (Rosenbaum, 2020), developmental and epileptic encephalopathies (Aledo-Serrano et al., 2020), and ST-segment-elevation myocardial infarction (Tam et al., 2020). The impact of this pandemic is important to be assessed in the rare disease population, a relatively vulnerable group of patients. Rare disease is defined to affect fewer than 5 per 10,000 people in the European population according to the World Health Organization (World Health Organization). The total number of rare diseases is estimated at 5000 to 8000. They are individually rare, but collectively rare diseases affect 6–8% of the European population (Simerka, 2009). In Hong Kong, one in 67 is living with at least one rare disease, representing 1.5% of the population (Chu et al., 2018). With majority of the rare diseases being chronically debilitating or life threatening, healthcare resource reallocation and public gathering restrictions during the pandemic may exacerbate the difficulties this population is facing and pose significant risk on patient's health and social well-being.

Increasing research have been done to assess the impact of the

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https://doi.org/10.1016/j.ejmg.2020.104062
Received 4 August 2020; Received in revised form 4 September 2020; Accepted 5 September 2020
Available online 12 September 2020
1769-7212/

https://doi.org/10.1016/j.ejmg.2020.104062

Covid-19 Pandemic in Indonesia

PeerJ

Full-length genome characterization and phylogenetic analysis of SARS-CoV-2 virus strains from Yogyakarta and Central Java, Indonesia

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Accepted 24 November 2020
Published 21 December 2020

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Additional Information and Declarations can be found on page 12

DOI 10.7717/peerj.10575

ABSTRACT

Background: Recently, SARS-CoV-2 virus with the D614G mutation has become a public concern due to rapid dissemination of this variant across many countries. Our study aims were (1) to report full-length genome sequences of SARS-CoV-2 collected from four COVID-19 patients in the Special Region of Yogyakarta and Central Java provinces, Indonesia; (2) to compare the clade distribution of full-length genome sequences from Indonesia ($n = 60$) from March to September 2020 and (3) to perform phylogenetic analysis of SARS-CoV-2 complete genomes from different countries, including Indonesia.

Methods: Whole genome sequencing (WGS) was performed using next-generation sequencing (NGS) applied in the Illumina MiSeq instrument. Full-length virus genomes were annotated using the reference genome of hCoV-19/Wuhan/Hu-1/2019 (NC_045512.2) and then visualized in UGENE v. 1.30. For phylogenetic

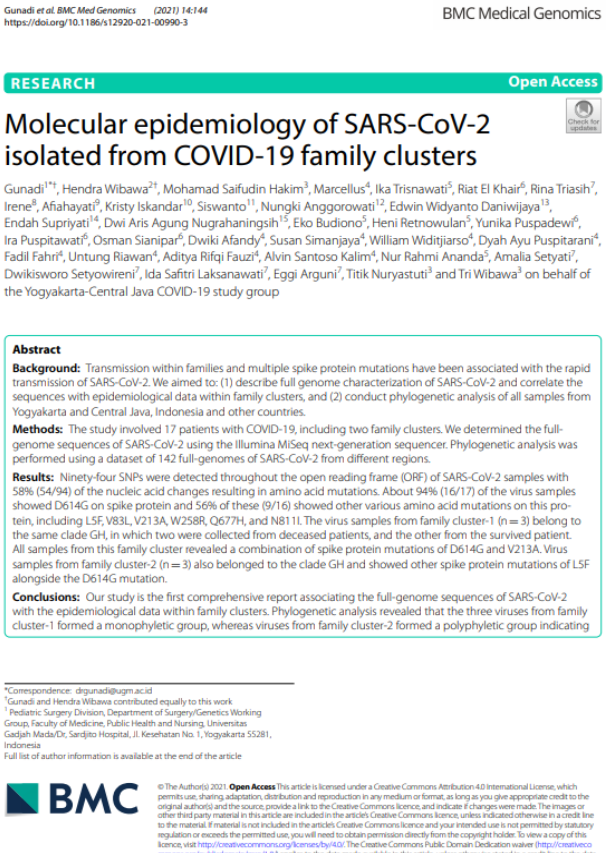
https://peerj.com/articles/10575/

Abstract

Background: Recently, SARS-CoV-2 virus with the D614G mutation has become a public concern due to rapid dissemination of this variant across many countries. Our study aims were (1) to report full-length genome sequences of SARS-CoV-2 collected from four COVID-19 patients in the Special Region of Yogyakarta and Central Java provinces, Indonesia; (2) to compare the clade distribution of full-length genome sequences from Indonesia (n = 60) from March to September 2020 and (3) to perform phylogenetic analysis of SARS-CoV-2 complete genomes from different countries, including Indonesia. **Methods**: Whole genome sequencing (WGS) was performed using next-generation sequencing (NGS) applied in the Illumina MiSeq instrument. Full-length virus genomes were annotated using the reference genome of hCoV-19/Wuhan/Hu-1/2019 (NC_045512.2) and then visualized in UGENE v. 1.30. For phylogenetic analysis, a dataset of 88 available SARS-CoV-2 complete genomes from different countries, including Indonesia, was retrieved from GISAID. **Results**: All patients were hospitalized with various severities of COVID-19. Phylogenetic analysis revealed that one and three virus samples belong to clade L and GH. These three clade GH virus samples (EPI_ISL_525492, EPI_ISL_516800 and EPI_ISL_516829) were not only located in a cluster with SARS-CoV-2 genomes from Asia but also those from Europe, whereas the clade L virus sample (EPI_ISL_516806) was located amongst SARS-CoV-2 genomes from Asia. Using full-length sequences available in the GISAID EpiCoV Database, 39 of 60 SARS-CoV-2 (65%) from Indonesia harbor the D614G mutation. **Conclusion**: These findings indicate that SARS-CoV-2 with the D614G mutation appears to become the major circulating virus in Indonesia, concurrent with the COVID-19 situation worldwide.

Abstract

Background: Transmission within families and multiple spike protein mutations have been associated with the rapid transmission of SARS-CoV-2. We aimed to: 1) describe full genome characterization of SARS-CoV-2 and correlate the sequences with epidemiological data within family clusters, and 2) conduct phylogenetic analysis of all samples from Yogyakarta and Central Java, Indonesia and other countries. **Methods**: The study involved 17 patients with COVID-19, including two family clusters. We determined the full-genome sequences of SARS-CoV-2 using the Illumina MiSeq next-generation sequencer. Phylogenetic analysis was performed using a dataset of 142 full-genomes of SARS-CoV-2 from different regions. **Results**: Ninety-four SNPs were detected throughout the open reading frame (ORF) of SARS-CoV-2 samples with 58% (54/94) of the nucleic acid changes resulting in amino acid mutations. About 94% (16/17) of the virus samples showed D614G on spike protein and 56% of these (9/16) showed other various amino acid mutations on this protein, including L5F, V83LI V213A, W258R, Q677H, and N811I. The virus samples from family cluster-1 (n=3) belong to the same clade GH, in which two were collected from deceased patients, and the other from the survived patient. All samples from this family cluster revealed a combination of spike protein mutations of D614G and V213A. Virus samples from family cluster-2 (n=3) also belonged to the clade GH and showed other spike protein mutations of L5F alongside the D614G mutation. **Conclusions**: Our study is the first comprehensive report associating the full-genome sequences of SARS-CoV-2 with the epidemiological data within family clusters. Phylogenetic analysis revealed that the three viruses from family cluster-1 formed a monophyletic group, whereas viruses from family cluster-2 formed a polyphyletic group indicating there is the possibility of different sources of infection. This study highlights how the same spike protein mutations among members of the same family might show different disease outcomes.



Gunadi et al. BMC Med Genomics (2021) 14:144
https://doi.org/10.1186/s12920-021-00990-3

BMC Medical Genomics

RESEARCH Open Access

Molecular epidemiology of SARS-CoV-2 isolated from COVID-19 family clusters

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Abstract

Background: Transmission within families and multiple spike protein mutations have been associated with the rapid transmission of SARS-CoV-2. We aimed to: (1) describe full genome characterization of SARS-CoV-2 and correlate the sequences with epidemiological data within family clusters, and (2) conduct phylogenetic analysis of all samples from Yogyakarta and Central Java, Indonesia and other countries.

Methods: The study involved 17 patients with COVID-19, including two family clusters. We determined the full-genome sequences of SARS-CoV-2 using the Illumina MiSeq next-generation sequencer. Phylogenetic analysis was performed using a dataset of 142 full-genomes of SARS-CoV-2 from different regions.

Results: Ninety-four SNPs were detected throughout the open reading frame (ORF) of SARS-CoV-2 samples with 58% (54/94) of the nucleic acid changes resulting in amino acid mutations. About 94% (16/17) of the virus samples showed D614G on spike protein and 56% of these (9/16) showed other various amino acid mutations on this protein, including L5F, V83L, V213A, W258R, Q677H, and N811I. The virus samples from family cluster-1 (n = 3) belong to the same clade GH, in which two were collected from deceased patients, and the other from the survived patient. All samples from this family cluster revealed a combination of spike protein mutations of D614G and V213A. Virus samples from family cluster-2 (n = 3) also belonged to the clade GH and showed other spike protein mutations of L5F alongside the D614G mutation.

Conclusions: Our study is the first comprehensive report associating the full-genome sequences of SARS-CoV-2 with the epidemiological data within family clusters. Phylogenetic analysis revealed that the three viruses from family cluster-1 formed a monophyletic group, whereas viruses from family cluster-2 formed a polyphyletic group indicating

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https://doi.org/10.1186/s12920-021-00990-3

Covid-19 Pandemic in Singapore

Do you remember the first time Covid-19 hit your consciousness? My first memory of it was right at the beginning of 2020 in a work meeting. Amongst the cheer of being in a new year and the anticipation of plans materializing, we were alerted to a situation developing and being asked to ensure that preparations were being made should things escalate. Without knowing what was to come, I appreciated that more than physical preparations being made, we were gearing up mentally. The then-unnamed virus turned out to be the challenge of our lifetime and the mental preparation was invaluable. It is still vital to keep hope alive in the days ahead.

In Singapore, the first case was reported on 23 January 2020. Almost 17 months later, as of the 17th of May, the country has seen 62,332 cases and 34 deaths.[1] Testing capabilities are in place and high risk individuals are routinely tested, with isolation and rigorous contact tracing conducted for positive cases. Vaccination programs for healthcare workers, seniors, students and other high risk individuals are underway and will be soon be available to the general population.[1]

Much of the information is available via the internet, so I wanted to tell some stories of people's lives in this season. More than 1 million people on our shores are considered foreigners but many more have family all over the world. Due to travel restrictions, people who have taken work overseas find themselves unable to meet their family for the foreseeable future. The separation takes its toll, especially for our front liners who give their all in caring and protecting their charges but are distanced from the ones who can refresh their spirits. Patients who already feel isolated by their rare conditions can sometimes feel even more distanced due to restrictions on social interactions. This was a stark reminder when one of our patients was critically ill. The family wished that he could be brought home to be surrounded and held by his extended family who had contributed to caring for him throughout his life and illness. Restrictions at that time meant that numbers of visitors would be severely restricted and even with a willing community, support was only available at arms' length.

Speaking of community, one of the bright sparks of hope is the concern shown amongst the members of this Society. As each of our own countries battled waves of infection, I have seen members offer encouragement, support and even physical help to each other. That in turn has inspired me to reach out to care for the next person. In between the more challenging periods, I am also reminded to look for opportunities to thrive. Since change is the only constant now, we have been able to try modes of clinical teaching and communication with patients that push our previous boundaries. Some ideas are successful and in others, we learn to do better. At the age of 95, my grandmother is one person who continues to push her boundaries, deciding to break her isolation safely by braving the last frontier - the internet. She has since declared YouTube to be the best thing ever. Jokes aside, for the first time, we have been able to gather as a family scattered over 5 countries to celebrate her birthday.

While movement is restricted, collaborations across institutions and borders are much easier. Without the need to be present at a physical location, we are now able to be far more inclusive than ever before. I have hope that when we are able to meet physically again, that these bonds forged through adversity will endure.

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1. www.gov.sg/features/covid-19

Breana Cham
Principal Genetic Counsellor
Genetics Service, Department of Paediatrics
KK Women's and Children's Hospital, Singapore

We would like to welcome the following new member of APSHG:

- Indra Lesmana (Indonesia)

How to be a member?

How to become an APSHG member?
Membership is open to those with keen interest in the study of human genetics, including basic, applied and medical genetics.

Categories of Membership:
Regular: Open to persons involved or interested in the study of human genetics in the Asia Pacific region with fully paid membership dues. Regular members have voting rights.

Junior: Open to persons under 30 years of age who are registered full-time students or trainees with fully paid membership dues. Junior members have no voting rights.

Life: Open to Regular members who have paid life membership dues. Life members have voting rights.

Senior: Open to members who have retired from full time employment. These persons should have been Regular Members and have paid annual membership dues for 5 years in total before being qualifying for Senior membership. They will then be entitled to the privilege of Regular members without fee. Senior members have voting rights.

Corporate: Upon recommendation of the Board, the Society may elect organisations that support the aims of the Society to Corporate membership. Each corporate member shall be entitled to nominate not more than two representatives to the Society and these nominees are subject to the approval of the Board. The Corporate members are not eligible to hold office or be members of the Board or to vote. Corporate members cannot use the name or logo of the Society for the purpose of advertisement or for any commercial or non-professional purposes. Corporate members shall seek in writing the written approval of the Board for use of the name or logo of the Society for any professional, education, or other matters. Corporate nominees are subject to similar restrictions imposed on the Corporate members.

Please register online: “Online Registration“

Source: https://www.apshg.info/memberships.html

Report from Malaysia on World DNA Day 2021 Celebration

World DNA Day is celebrated every year on 25th April to honour the achievement of the Human Genome Project (HGP) which was completed in 2003 and the ground-breaking elucidation of the model structure of DNA double helix which was published in Nature magazine on 25 April 1953. After the United States Congress passed simultaneous resolutions designating April 25th as DNA Day, the National Human Genome Research Institute (NHGRI) began celebrating it every year on April 25th.

The main aim of World DNA Day is to provide scholars, educators, and the general public with a chance to study about and cherish the most current developments in genetic scientific research as well as to spread awareness on how DNA affect our lives. From January to May of each year, the NHGRI also invites communities to organize DNA Day celebrations.

The other reasons why the World DNA Day was important was because it recognized advances in science research. On World DNA Day, we honoured the contributions that contributed to the awareness we had today, as well as the ongoing studies that would contribute to tomorrow's achievements. It also inspired people to discover more about their genetic heritage. The science that had led to the celebration of World DNA Day was the science that connected us to our roots. This day contributed to our sense of identity by inspiring us to explore further into discovering who we are and where we came from. In addition, it was the day on which the general population could hear more about genetics and genomics. There was a lot to understand about the structure and work of genetics, from human origins to gene editing. On World DNA Day, the public was urged to seek out any and all possible facts in order to understand further about their genetic structure and the molecular biology of all living organisms.

In Malaysia, we celebrated World DNA Day 2021 virtually on 25 April 2021.This programme was organised by the Malaysian Society of Human Genetic (MSHG) in collaboration with the Malaysian Human Variome Project (MyHVP), Universiti Malaysia Sarawak (UniMAS), Faculty of Medicine and Health Sciences, as well as several departments at Universiti Sains Malaysia (USM) Health Campus including the Human Genome Center, School of Medical Sciences, USM, Department of Psychiatry, School of Medical Sciences, USM, School of Health Sciences, USM which successfully organized this program. There were 130 participants involved including students and teachers from different schools, parents and patients' guardians, professionals, medical students and general public. This programme started at 9.30 am until 12.45 pm. There were a few interesting topics discussed in this programme which are firstly, talk on The Beauty of Genetic Science: Get to Know your DNA by Assoc. Prof. Dr Sarina Sulong from USM, secondly, talk on Work in Areas of Human Genetics by Dr Muhammad Hamdi Mahmood from UNIMAS, thirdly, Fetal Genetic Disorders and Abortion in Islam by Dr Mujahid Bakar from USM and lastly, Genetic Diseases and Psychological Effects by Dr Norzila Zakaria from USM. This activity could develop awareness among people in Malaysia about the importance of our DNA in health and how it can transform our future.

This virtual programme succeed in achieving their objectives which were increasing knowledge of the importance of DNA as the basic unit of human inheritance, providing exposure to careers in allied health sciences, increasing knowledge on the importance of mental health in dealing with diseases of a congenital nature, and assisting the population, especially parents, in managing genetic patients and also introducing the MSHG as an organization concerned with the health and welfare of genetic patients. this seminar, which was successful with the collaboration and support of all parties, can benefit all levels of society with the sharing of knowledge and information. This in turn can help provide awareness of DNA and genetic diseases as well as mental health management not only in the family but the patient himself.

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Chung et al. Orphanet J Rare Dis (2021) 16:119
https://doi.org/10.1186/s13023-021-01766-9

Orphanet Journal of Rare Diseases

RESEARCH Open Access

A thematic study: impact of COVID-19 pandemic on rare disease organisations and patients across ten jurisdictions in the Asia Pacific region

Claudia Ching Yan Chung[1], Yvette Nga Chung Ng[1], Ritu Jain[2,3*] and Brian Hon Yin Chung[1*]

Abstract

Background: This study assesses the areas and extent of impact of the Coronavirus Disease of 2019 (COVID-19) pandemic on rare disease (RD) organisations in the Asia Pacific region. There is no existing literature that focuses on such impact on RD organisations in any jurisdictions, nor RD populations across multiple jurisdictions in the Asia Pacific region. A cross-sectional survey was distributed to RD organisations between April and May 2020. Quantitative and qualitative data on the impact of COVID-19 on RD organisations and patients were collected from the organisation representative's perspective. Qualitative data was analysed using thematic analysis. A follow-up focus group meeting was conducted in August 2020 to validate the survey findings and to discuss specific needs, support and recommendations for sustainable healthcare systems during the pandemic.

Results: A total of 80 RD organisations from Australia, Hong Kong Special Administrative Region of China, India, Japan, mainland China, Malaysia, New Zealand, the Philippines, Singapore and Taiwan participated in the study. Of all, 89% were concerned about the impact of pandemic on their organisations. Results indicate that 63% of the organisations functioned at a reduced capacity and 42% stated a decrease in funding as their biggest challenge. Overall, 95% believed their patients were impacted, particularly in healthcare access, social lives, physical health, psychological health and financial impact. Specifically, 43% identified the reduced healthcare access as their top impact, followed by 26% about the impact on daily living and social life. Focus group meeting discussed differential impact across jurisdictions and point towards telemedicine and digitalisation as potential solutions.

Conclusions: This serves as the first study to assess the impact of COVID-19 on RD patients and organisations across multiple jurisdictions in the Asia Pacific region, identifying major themes on the impact on both RD patients and organisations. By including 80 organisations from ten jurisdictions, our study presents the most comprehensive assessment of the pandemic's impact to date. It highlights the need for mental health support and sheds light on moving towards telemedicine and digitalisation of organisation operation, which constitutes a sustainable model in times of pandemics and beyond.

Keywords: COVID-19, Rare disease, Rare disease organisation, Rare disease patients, Asia Pacific, Thematic analysis

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Introduction

Since the outbreak of the Coronavirus Disease of 2019 (COVID-19) in Wuhan, China in December 2019, the pandemic had spread to many countries throughout the world. The outbreak was declared a public health

https://doi.org/10.1186/s13023-021-01766-9

Intractable & Rare Diseases Research. 2021; 10(1):11-16. 11

Review

DOI: 10.5582/irdr.2020.03101

Surveillance and prevalence of fragile X syndrome in Indonesia

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SUMMARY Fragile X syndrome (FXS) is the most prevalent inherited cause of intellectual disability (ID) and autism spectrum disorder (ASD). Many studies have been conducted over the years, however, in Indonesia there is relatively less knowledge on the prevalence of FXS. We reviewed all studies involving FXS screening and cascade testing of the high-risk population in Indonesia for two decades, to elucidate the prevalence, as well as explore the presence of genetic clusters of FXS in Indonesia. The prevalence of FXS in the ID population of Indonesia ranged between 0.9-1.9%, while in the ASD population, the percentage was higher (6.15%). A screening and cascade testing conducted in a small village on Java Island showed a high prevalence of 45% in the ID population, suggesting a genetic cluster. The common ancestry of all affected individuals was suggestive of a founder effect in the region. Routine screening and subsequent cascade testing are essential, especially in cases of ID and ASD of unknown etiology in Indonesia.

Keywords fragile X syndrome, intellectual disability, genetic screening, cascade testing

1. Introduction

Fragile X syndrome (FXS) is an X-linked inherited condition that causes developmental problems, including intellectual disability (ID). FXS is caused by the expansion of the cytosine-guanine-guanine (CGG) trinucleotide repeat in the 5' untranslated region (UTR) of the fragile X mental retardation (*FMR1*) gene (OMIM 309550). It has a prevalence of 0.5 to 3 percent in different populations with intellectual disability (ID) and autism spectrum disorder (ASD) (*1*). FXS is characterized by ID and emotional and behavioral disorders, including a short attention span, hyperactivity, tactile defensiveness, and poor eye contact (*2*). Dysmorphic clinical features of FXS include large and prominent ears, macroorchidism during and after puberty, single palmar crease, and hyperextensible joints (*3*). About 50 to 60% of male patients with FXS also have features of autism spectrum disorder (ASD), and FXS is considered to be the most common single gene cause of ASD (*4*). The expansion of CGG trinucleotide repeats is unstable within a specific threshold, with variable length in the normal population. The range of repeats in a normal individual is 5 to 44 repeats. Individuals with 45-54 repeats have variable expansion characteristics and this allele is called an intermediate or 'gray zone' allele, while individuals with 55-200 repeats are classified as premutation carriers. The phenotypes in FXS are associated with more than 200 CGG repeats and methylation, and this range is called the full mutation. Expansion instability usually results from maternal transmission, however, 1 or 2 AGG anchors after every 10 CGG repeats can lead to less frequent expansion to the full mutation when passed on by a mother to the next generation (*5*).

The first cytogenetic analysis of FXS identified the fragile site on the long arm of chromosome X located at Xq27.3, whereby the syndrome was named (*6*). Further molecular analysis for diagnosis of FXS, including a polymerase chain reaction (PCR) based method was introduced after the *FMR1* gene molecular structure was identified in 1991 (*7*). There have been many PCR protocols developed to measure the size of CGG repeats, and PCR is one of the most inexpensive and convenient methods for diagnosis. However, the DNA fragment of expanded repeats in the mid-high premutation range does not amplify well in PCR, so full mutation alleles cannot be detected. Consequently, other methods are

https://doi.org/10.5582/irdr.2020.03101

Prospective Clinical Research Report

Journal of INTERNATIONAL MEDICAL RESEARCH

Journal of International Medical Research
49(2) 1–10

DOI: 10.1177/0300060520987789
journals.sagepub.com/home/imr
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Effect of semaphorin 3C gene variants in multifactorial Hirschsprung disease

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Abstract

Objective: Cluster genes, specifically the class 3 semaphorins (*SEMA3*) including *SEMA3C*, have been associated with the development of Hirschsprung disease (HSCR) in Caucasian populations. We aimed to screen for rare and common variants in *SEMA3C* in Indonesian patients with HSCR.
Methods: In this prospective clinical study, we analyzed *SEMA3C* gene variants in 55 patients with HSCR through DNA sequencing and bioinformatics analyses.
Results: Two variants in *SEMA3C* were found: p.Val337Met (rs1527482) and p.Val579= (rs2272351). The rare variant rs1527482 (A) was significantly overrepresented in our HSCR patients (9.1%) compared with South Asian controls in the 1000 Genomes (4.7%) and Exome Aggregation Consortium (ExAC; 3.5%) databases. Our analysis using bioinformatics tools predicted this variant to be evolutionarily conserved and damaging to SEMA3C protein function. Although the frequency of the other variant, rs2272351 (G), also differed significantly in Indonesian patients with HSCR (27.3%) from that in South Asian controls in 1000 Genomes (6.2%) and ExAC (4.6%), it is a synonymous variant and not likely to affect protein function.

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https://doi.org/10.1177%2F0300060520987789

Intractable & Rare Diseases Research **Advance Publication** P1

Case Report

Advance Publication DOI: 10.5582/irdr.2020.03143

Clinical manifestation and genetic analysis of familial rare disease genodermatosis xeroderma pigmentosum

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SUMMARY Xeroderma pigmentosum (XP) is a rare autosomal recessive disease characterized by hypersensitivity of the skin to ultraviolet radiation and other carcinogenic agents. This ailment is characterized by increased photosensitivity, skin xerosis, early skin aging, actinic keratosis, erythematous lesions, and hyperpigmentation macules. In this serial case report, we presented four cases with XP from two families in Indonesia. Both families were referred from rural referral health centers, and each family has two affected siblings. They had freckle-like pigmentation on the face, trunk, and extremities, which progressed since childhood. One patient of family 2 died because of an infectious disease. Histopathological examination using cytokeratine (CK), CD10, and Ber-EP4 staining from available tissue biopsy of one affected case of family 1 identified basal cell carcinoma (BCC) on the cheek and melanoma on the right eye. Mutation analysis found *ERCC2*, c2047C>T and *XPC*, c1941T>A in the first and second families, respectively. We suppose that this is the first case report of XP in Indonesia that incorporates clinical examination, genetic analysis, and extensive histopathological examination, including immunohistochemistry staining, and a novel pathogenic variant of *XPC* was found in the second family.

Keywords xeroderma pigmentosum, familial, autosomal recessive, photosensitivity, XPC, mutation

1. Introduction

Xeroderma pigmentosum (XP) is characterized by increased photosensitivity, skin xerosis, early skin aging, actinic keratosis, erythematous lesions, and hyperpigmentation macules (*1,2*). Another feature is abnormal lentiginosis (freckle-like pigmentation due to increased numbers of melanocytes) on sun-exposed areas. This is followed by areas of increased or decreased pigmentation, skin aging, and multiple skin cancers if the individuals are not protected from sunlight (*3*). These manifestations are due to cellular hypersensitivity to ultraviolet (UV) radiation resulting from a defect in DNA repair. The mutation that causes XP affects one of eight XP-related genes, including *XPA*, *XPB*, *XPC*, *XPD*, *XPE*, *XPF*, *XPG*, and *XPV* (XP variant), which encodes the *nucleotide excision repair* (NER) mechanism (*4,5*).

XP cases can be found in almost every place in the world with variable prevalence (*6,7*). In the United States, this condition affects one person per one million population (*6,8*). In Europe, this case affects up to 2.3 persons per one million live births. In the Middle East, the prevalence of XP is around 15-20 persons per one million population (*6-8*). Consanguinity itself is highly related to the incidence of XP. Therefore, XP cases are higher in areas where consanguinity is common, including North Africa and the Middle East. Consanguinity is an important factor in autosomal recessive disorders (*8*). Up to 92.8% of XP patients in Libya had consanguinity history (*7*).

XP is a rare autosomal recessive inherited genodermatosis with only a few cases worldwide. Most people are not aware of this condition, which is why XP patients are often neglected and do not receive medical assistance. Seven different genes (labeled A to G), which have deficient excision repair of ultraviolet radiation-

https://doi.org/10.5582/irdr.2020.03143

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PSGCA Facts and Figures 2020
- 31 members
- 2 webinars with 293 attendees
- 8 virtual Board of Directors meetings
- 13 external events promoted

What's New!
The Professional Society of Genetics Counselors in Asia (PSGCA) co-hosted webinars with the Institute of Human Genetics, University of the Philippines, Manila to engage genetic counselors and genetics health professionals with ongoing discussions towards professional development in the Asia-Pacific region.

The first webinar focused on Peer Supervision in Genetic Counseling was held on 17 December 2020. The resource speaker was Niby Elackatt, MSc, the co-founder and director of genetic counseling, Bluegene Healthtech (India) and she highlighted the importance of peer supervision as a strategy to continuously develop competencies in genetic counseling through clinical conversations and discussing challenging cases. This webinar was attended by 103 participants from the Asia-Pacific region.

Held on 23 February 2021, the 2nd webinar focused on Genetics Education for Health Professionals. Emily Edelman, MS, CGC of the Jackson Laboratory (USA) discussed the approaches and resources for genetics education that can be used by health professionals as they integrate genetics and genomics in their clinical practice. This webinar was attended by 223 participants from the Asia-Pacific region.

Future webinar topics, as recommended by webinar attendees, include: patient communication and counseling, patient management based on genomic data, and genetic test selection.

In addition, the PSGCA collaborated with iBRCAf and American College of Medical Genetics and Genomics in promoting their annual events. They also have offered special registration rates to our members.

5th INTERNATIONAL BRCA FORUM VIRTUAL
iBRCAf
BRCA & Beyond

ACMG
American College of Medical Genetics and Genomics

Thank you for the support. We will keep our members notified on future events.

Call To Action:
Would you like to facilitate or mentor a challenge case discussion? Do you have a challenging case that we can support you with?

The PSGCA is planning a 'Challenging Case Discussions' workshop for genetic counselors who would like to elevate their facilitation, mentorship and public speaking skills. This workshop offers an ideal forum for discussion of cases which have challenged genetic counselors and stimulated considerable reflection.

We are currently looking for certified or experienced genetic counselors who are willing to share their experience and knowledge in helping others manage challenging cases. If you're interested, please email Ms. Juliana Lee juliana@

psgca.org by stating your name, country, area of specialty, years of practice and how you wish to contribute to this activity.
Interested to participate in PSGCA events? Become one of our members.

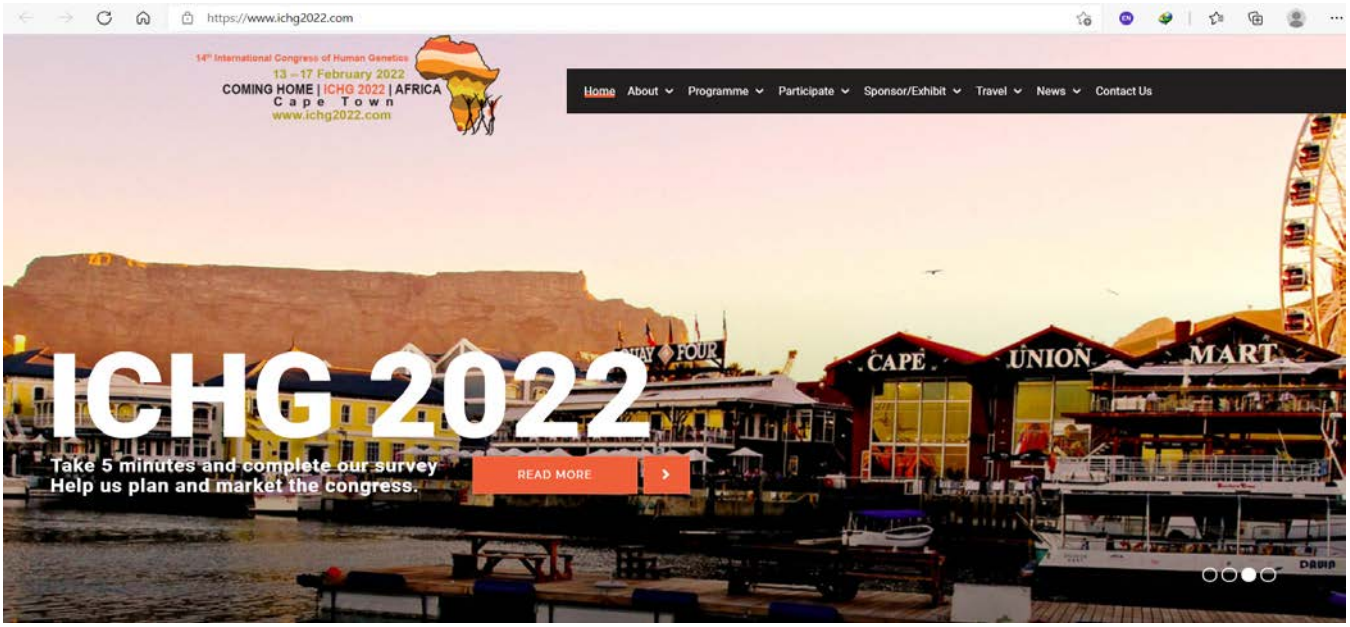
How to become a PSGCA member?
PSGCA membership is open to genetic counselors, genetic nurses and students in genetic counseling programs in the Asia Pacific region. The membership fee is only SGD $10 per year.

As PSGCA is a special interest group of APSHG, one must first become a member of the APSHG. You can register for both societies' membership at https://www.apshg.info/memberships.html.

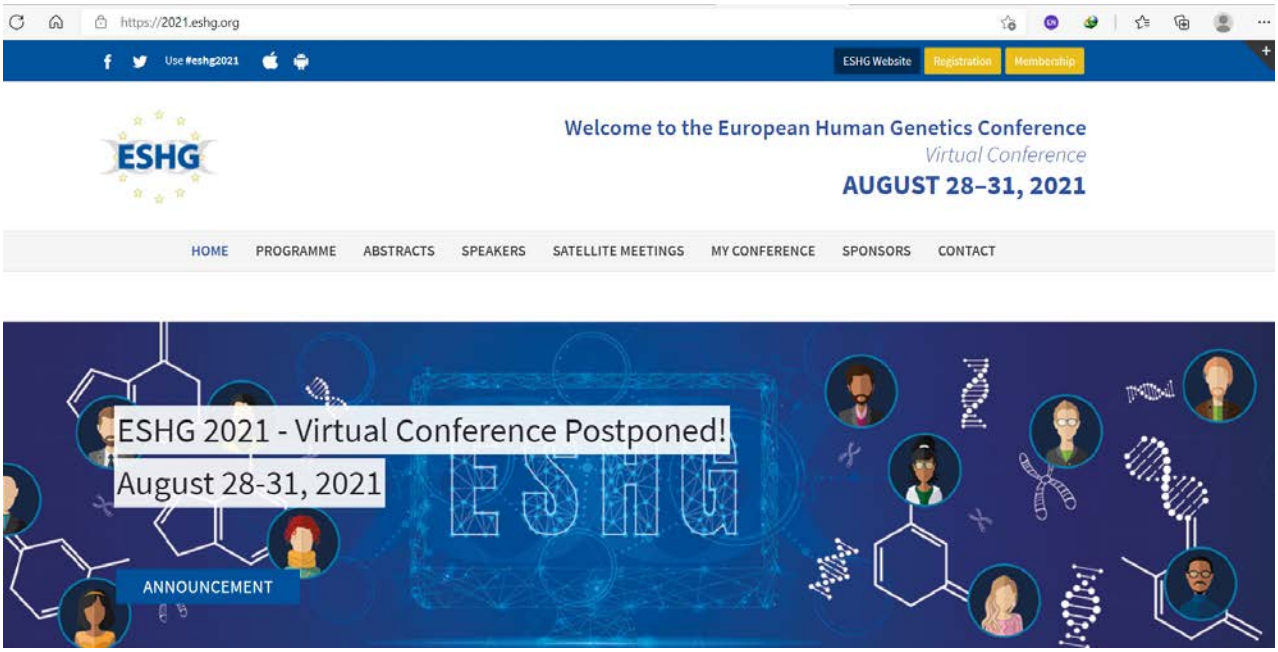
Have you renewed your membership?
A reminder email was sent to all PSGCA members of 2020 for membership renewal. If you have not received or have any questions, please email admin@psgca.org

Upcoming Events

- **14th International Congress of Human Genetics (ICHG2022), Cape Town, South Africa , 9 - 10 March 2022.**



- **European Human Genetics Conference, Virtual Conference, Auugust 28-31, 2021**



- **Board of Genetic Counseling - India 6th Annual International Conference, July 2-4, 2021**

- **Variant Effect Prediction Training Course, Virtual Course, 21-23 September 2021**

- **The 2nd Indonesian Society of Human Genetics (InaSHG) Conference, September 4-5 2021**

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InaSHG
INDONESIAN SOCIETY OF HUMAN GENTICS

1ST ANNOUNCEMENT

The 2nd Indonesian Society of Human Genetics (InaSHG) Conference

"Research and Clinical Practice in Genomic Era : Challenges and Opportunities"

2021 SEPTEMBER 04-05
EXCLUSIVE VIRTUAL CONFERENCE

LIST OF SPEAKERS

INTERNATIONAL SPEAKERS :

1. Brian Hon-Yin Chung, MD, Ph.D (The University of Hong Kong, Hong Kong)
2. Erik Sistermans, Ph.D (VU University Medical Centre, Netherlands)
3. Gerard Pals, Prof., Ph.D (VU University Medical Centre, Netherlands)
4. Ghada El-Kamah, Prof., MD, Ph.D (Department of Clinical Genetics, National Research Center, Egypt)
5. Helger Yntema, Ph.D (Radboud University Nijmegen Medical Centre, Netherlands)
6. Juliana M.H Lee, FHGSA (Genetic Counselling) (Genetic Counselling Asia)
7. Lai Poh San, Prof., MD, Ph.D (National University of Singapore, Singapore)
8. Sarina Sulong, Ph.D (Universiti Sains Malaysia, Malaysia)
9. Thong Meow Keong, Prof., MD, Ph.D, FHGSA (Genetic Counselling) (Department of Clinical Genetics, University of Malaya, Malaysia)
10. Zilfalil Alwi, Prof., MD, Ph.D (Universiti Sains Malaysia, Malaysia)

INDONESIAN SPEAKERS :

1. Achmad Zulfa Juniarto, MD, Ph.D, Sp.And (Center for Biomedical Research, UniversitasDiponegoro)
2. Agustini Utari, MD, Ph.D, Sp.A (K) (Center for Biomedical Research, Universitas Diponegoro)
3. Ahmad Fariz Malvi Zamzam Zein, MD, Sp.PD (Waled Hospital Cirebon-Universitas Swadaya Gunung Jati)
4. Akhmad Makhmudi, MD, Ph.D, Sp.BA(K) (Sardjito Hospital-Universitas Gadjah Mada)
5. Aru Wisaksono Sudoyo, Prof., MD, Ph.D, Sp.PD-KHOM, FACP (Universitas Indonesia)
6. Cita Rosita Sigit Prakoeswa, Prof. MD, Ph.D, Sp.KK(K), FINSDV (Universitas Airlangga)
7. Damajanti Rusli Sjarief, Prof., MD, Ph.D, Sp.A(K) (Cipto Mangunkusumo Hospital-Universitas Indonesia)
8. Gara Samara Brajadenta, MD, Ph.D (Universitas Swadaya Gunung Jati)
9. Gunadi, MD, Ph.D, Sp.BA (Sardjito Hospital-Universitas Gadjah Mada)
10. Herawati Sudoyo, Prof., MD, Ph.D (Eijkman Institute for Molecular Biology)
11. Iswari Setianingsih, MD, Ph.D (Eijkman Institute for Molecular Biology)
12. Mohamad Erwin Indrakusuma, MD, Sp. MK (Waled Hospital Cirebon-Universitas Swadaya Gunung Jati)
13. Muhammad Hussein Gasem, Prof., MD, Ph.D, Sp.PD-KPTI (Universitas Swadaya Gunung Jati)
14. Ramdan Panigoro, Prof., MD, Ph.D (Universitas Padjadjaran)
15. Samuel Johny Haryono, MD, Ph.D, Sp.B K(Onk) (Dharmais Cancer Hospital)
16. Sofia Mubarika Haryana, Prof., MD, Ph.D (Universitas Gadjah Mada)
17. Sultana MH Faradz, Prof., MD, Ph.D (Center for Biomedical Research, Universitas Diponegoro)
18. Tri Indah Winarni, Prof., MD, Ph.D (Center for Biomedical Research, Universitas Diponegoro)
19. Tri Wibawa, Prof., MD, Ph.D, Sp.MK (Universitas Gadjah Mada)
20. Yunia Sribudiani, Ph.D (Universitas Padjadjaran)

REGISTRATION FEE

Participant	Domestic	Overseas
Presenter	IDR 300.000	USD 20
Non-Presenter	IDR 200.000	USD 15

PUBLICATION OPPORTUNITIES

- International Journal indexed by Scopus
- National Journal accredited by Indonesian Higher Education Commission (SINTA 1-3)

CALL FOR ABSTRACT

Abstracts are invited on all aspects of health related research, especially the conference sub-themes below:

1. Molecular Genetics and Techniques
2. Counseling Genetics and Social Aspects
3. Clinical Genetics
4. Immunology
5. Communicable Diseases
6. Non-Communicable Diseases
7. Health Technology and Medical Treatment
8. Pharmaceutical Sciences
9. Health Promotion, Health Policy and Education
10. Mental Health
11. Environmental and Occupational Health
12. Sport Sciences and Physical Education
13. Cancer Biology and Cancer Therapeutics
14. Others

CONFERENCE TOPICS

- Inborn Errors of Metabolism and Mendelian Diseases
- Disorder of Sex Development: From Genetics to Management
- Hereditary Cancer and Genetics
- Congenital Anomalies and Complex Genetic Disorders
- Challenge on Diagnosis and Management of Hemoglobinopathies
- Recent Advances in Molecular Genetics and Genomic Diagnostic
- Molecular Biology, Infection and Medical Ethic

IMPORTANT DATES

ACTIVITIES	DATES
Abstract Submission Deadline	August 23, 2021
Notification of Abstract Acceptance	August 27, 2021
Registration Deadline	September 03, 2021
Conference	September 04 - 05, 2021

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