



**Report on Global
Newborn
Screening
IFCC/ISNS Task**

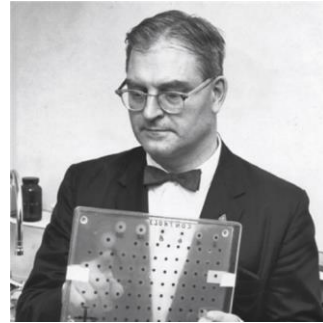
**October 30th, 2022
Drs. James Bonham (UK) and Van Leung-Pineda (USA)
Brussels, Belgium**



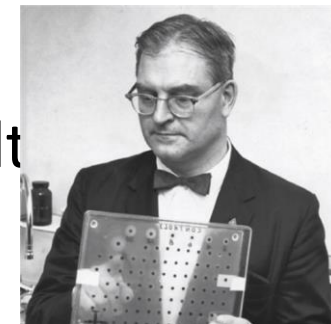
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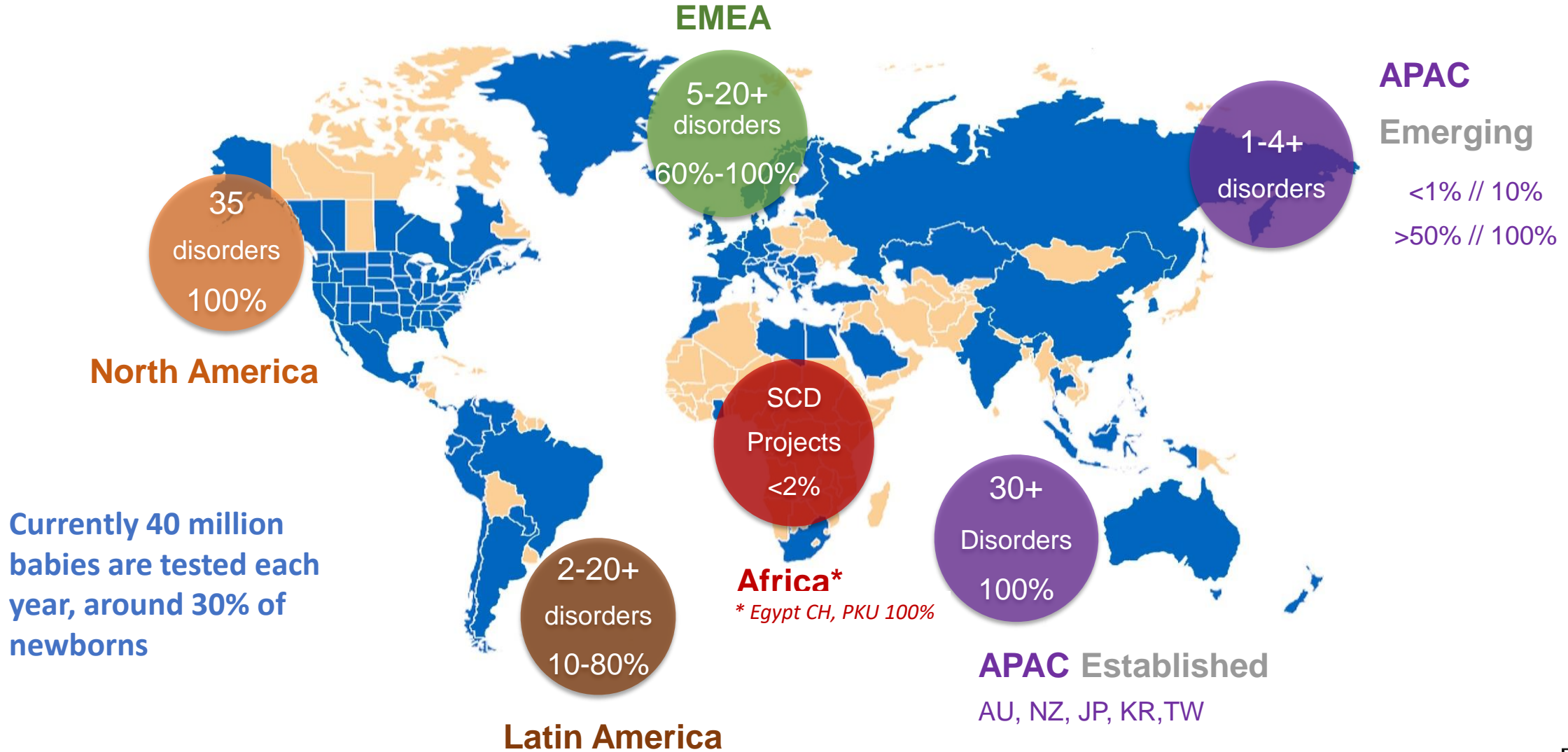
- Newborn Screening (NBS) started in the 60s with Dr. Robert Guthrie's pioneering work on phenylketonuria
- Approximately 750 m babies have now been tested and 60,000 benefitted from life changing treatment for PKU alone
- He gave us a test but perhaps more importantly a simple means of sample collection
- Quickly progressed to include other disorders
- Cost effective for the Healthcare System-detect and treat before irreversible symptoms affect the patient
- “One of the major Public Health Advances of the 20th Century”



- Of course as in most of medicine, there is a balance and sometimes difficult choices to make
- The patients/families believe themselves to be well and this gives us a particular burden of responsibility
- “All screening programmes do harm; some do good as well, and, of these, some do more good than harm....” *Gray, BMJ (2008) 336:480*
- More screening does not mean better screening
- Screening which is well organized and delivered as a carefully monitored programme linked to structured treatment is most effective and brings most success



World coverage and the challenge ahead



Task Force, Objective and Strategy

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- Started in 2021
 - 11 Members and 15 Corresponding members
 - <https://www.ifcc.org/executive-board-and-council/eb-task-forces/task-force-on-global-newborn-screening/>
- Objective: To help develop appropriate screening programmes worldwide including low/middle income countries
- First step: To raise awareness of the potential offered by appropriate newborn screening – helped by the formation of the Task Force and presentation of educational Webinars
- Second step: Conduct a worldwide survey
- Third step: Identify candidate regions/countries who may benefit most from support in the short term
- Fourth step: Arrange in country meetings involving local advocates, international support, key national health policy makers
- Fifth step: Plan pilot activity



Task Force: Guidelines for progress

- When planning, it is important to remember that ***'more screening does not automatically mean better screening'***
- Rather newborn screening that is well organized and appropriate to the local need with access to effective treatment, offers the best promise of success
- There are probably a few key messages when planning for success and sustainability:
 - Work with local advocates to ensure that screening is 'home grown and home owned'
 - Start small with relatively common disorders that are easy to test and inexpensive to treat
 - Engage with community groups, parents, physicians and health care policy makers
 - Remember that newborn screening is not just a test
 - Undertake careful planning that recognizes the whole pathway from first contact with the family, through to taking a sample, transport, analysis, reporting results, ensuring that affected children receive treatment
 - Continually monitor and assess the integrity of the pathway
 - Invite regular scrutiny
 - Produce annual reports and review progress



- **IFCC** Related Divisions within IFCC and the wider global IFCC community
- **AACC** (American Association of Clinical Chemistry)
 - NBS Symposiums and Workshops in Colombia (2017) in Cali, Morocco (2019) in Marrakesh, India (2019) in Goa, Planned workshop in Zambia in collaboration with AFCC (African Federation of Clinical Chemistry)
- **ASH** (American Society of Hematology)
 - Has a Sickle Cell Disease identification and intervention program in Africa named CONSA (Consortium on Newborn Screening in Africa). Includes Ghana, Kenya, Liberia, Nigeria, Tanzania, Uganda and Zambia currently. Have some infrastructure in these countries for NBS
- **CLSI** (Clinical and Laboratory Standards Institute) - can provide Guidelines and other educational materials (English)
- **APHL** (Association of Public Health Laboratories)
- **CDC** (Centers for Disease Control and Prevention) - can provide training materials and host and train individuals
- **SLEIMPN** (Latin American Society of Inborn Errors of Metabolism and Newborn Screening) - can help identify potential candidates for Latin America
- **WHO** (World Health Organisation) have expressed an interest in the inclusion of basic newborn screening as an important part of neonatal care and may be present at 'in country' meetings.

IFCC TF-NBS Survey

August 2021

- Survey was live in April 2021 and closed June 30 2021
- Was posted on IFCC TF website and distributed to different mailing lists
 - IFCC members
 - Commercial newsletters
 - Local societies
- Received approximately 425 responses from 84 countries

- In Countries with NBS, the top 4 disorders that were screened for were: CH, PKU, CAH and CF
- Most respondents (93%) would find a partnership with IFCC-ISNS beneficial
 - By providing support with development of education materials and guidelines
 - by training of laboratory, follow up and clinical personnel
 - by providing Laboratory Review/Quality Audit
- 100% reported interest working with TF to establish NBS (34 responses). 56% of these (19) want to be contacted with more details

- Local multidisciplinary involvement with key opinion leaders as advocates
- Local government interest
- Existing infrastructure to support sample taking and transport
- Access to treatment
- Attainable gains in the Task Force's timeframe

- Africa
 - South Africa
 - Nigeria
- South America
 - Bolivia
- Eastern Europe
 - Contact after last TF meeting

South Africa

- National Health Laboratory Service and University of Cape Town – **Public**
 - Screening is very **limited** in South Africa and serves approximately **10% of the population**, no screening is available for the majority of the population
 - CH screening is performed on **cord blood** in approximately 36000 newborns per year that fall into the drainage area of the Red Cross Children's Hospital in Cape Town, South Africa but this may not continue
 - The only other screening is **expanded NBS offered to babies born in the private healthcare sector.**
- Followed up in early September
 - There are enthusiastic local advocates
 - There are excellent transport and confirmatory testing arrangements in place
 - There is access to treatment
 - There is an interest at governmental level to consider nationwide screening at least for congenital hypothyroidism
 - There is interest from WHO
- The plan is to have an 'in country' meeting in February 2023 and follow-up at the WHO Maternal and Child meeting in May 2023 in Cape Town



Nigeria

- There is enthusiasm in the longer term to offer universal screening but there is a real opportunity to begin with the introduction of screening for CHT and Sickle in UCH births (24000 pa) with the aim of pushing to tertiary, secondary, primary facility and community births in time.
- IT connectivity available both in the Lab and in other aspects of the screening pathway
- John Anetor has been contacted and asked to outline a proposal for an in country meeting.
- It is hoped that the meeting will include: governmental representation, health policy makers, local advocates, clinicians and will include representation from WHO if this can be organized.
- It is hoped that a meeting could be arranged early in 2023.



Latin America

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- Reached to local expert Dr. Gustavo Borrajo (Marta Ascurra, Cecilia Queijo, Van Pineda)
- Discussed 4 possible candidates with promising local support to obtain more information from
 - Bolivia: 2 survey responses
 - Peru: 1 survey response
 - DR: 2 responses
 - Guatemala: 4 responses
- Candidates have some infrastructure but in need of a push
- Follow up survey received responses from Bolivia and Guatemala
- Bolivia has limited screening in some regions but this lacks co-ordination and coverage
- There is interest to form a coherent national program with government recognition
- Initiated contact with local Health Ministry representative and local endocrinology and IFCC representatives
- Confirmed receipt. Representative requested official note to Bolivia's Health Minister to initiate a technical project report
- Note to Health Minister was sent

- While in most of Europe national newborn screening programmes are in place there are marked differences in the number of conditions included ranging from 2 – 48
- In Eastern Europe in particular coverage for the population can be poor and testing very limited. Nevertheless, there are clear advocates in many of these countries and working with partner countries in western Europe there is the real possibility of development.
- Those countries that are working on this include:
 - Romania
 - Georgia
 - Ukraine
 - Albania
- MetabERN (a European Commission supported rare disease reference network) have expressed interest to help support development and consider routes to confirmatory testing and treatment
- There is a meeting planned for 28th and 29th Nov 2022 in Frankfurt to discuss

- Move from the exploratory phase to local visits
- Short-medium term
 - Engage the local IFCC societies
 - Eastern Europe, Planned for Nov 2022
 - South Africa, planned visit in Feb 2023, possible follow up in May
 - Bolivia, will plan initial visit early 2023, and schedule follow up when on site
 - Continue communication with Nigeria in 2023
- Build for the future
 - Other positive responses from the survey process, i.e. Uganda, Dominican Republic, Nepal

A note about International Neonatal Screening Day

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- Dr. Robert Guthrie's birthday in 28th June 1916
- ISNS, IPOPI (International Patient Organization for Primary Immunodeficiencies), ESID (European Society for Primary Immunodeficiencies) sponsored awareness and education about Newborn Screening and its positive impact and promote advancement
- A lot of partner organisations around the world
- First celebrated in 2021, then 2022
- Awareness raining but already some significant impacts in 2022
- Looking forward to INSD 2023!

Pre Task Force Webinar: Expanding Newborn Screening Globally

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 **IFCC Live Webinar on**
Expanding Newborn Screening Globally:
Reducing Infant Mortality Through Early
Diagnosis - 4th November, 2020

Piero Rinaldo (US) Professor of Laboratory Medicine and Pediatrics, Co-director Biochemical Genetics Laboratory, Mayo Clinic, Rochester MN	Jim Bonham (UK) President International Society of Neonatal Screening, National Newborn Screening Laboratory Lead	Pranesh Chakraborty (CA) Executive Director & Chief Medical Officer, Ontario Newborn Screening Program
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Collaborative Laboratory Integrated Reports (CLIR): A global IT platform to achieve and sustain precision newborn screening	Developing Newborn Screening in Low and Middle Income Countries	Newborn Screening: Beyond the test
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Registration link (Scan QR code)

<https://www.workcast.com/register?cpak=5982974887417799>

Date: 4 November 2020
Time: 9 AM (Eastern Standard), 3 PM (European), 10 PM (Beijing)



Chair: Khosrow Adeli (Canada) Professor of Clinical Biochemistry, University of Toronto, President IFCC

IFCC Webinars

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Live Series

2022

www.ifcc.org



IFCC Live Webinar on Aspects of Newborn Screening



Moderator



Dr. Van Leung-Pineda
[USA]
Children's Healthcare of Atlanta,
Emory University School of
Medicine

Newborn Screening in the
Genomics Era



Prof. James Robert Bonham
[UK]
Sheffield Children's NHS FT

Developing and maintaining a
national newborn screening
programme in a low-middle income
country



Dr. Carmencita Padilla
[Philippines]
University of the Philippines Manila

Laboratory Aspects of Testing
on Dried Blood Spots



Dr. Dianne Webster
[NZ]
NZ Newborn Screening Programme,
Auckland City Hospital

Date: Oct 5, 2022

Time: 8 AM (Eastern Standard), 3 PM (Central European), 9 PM (Beijing)



Thank you for your attention and support

For further information, visit
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