Report: Visit to the Pediatric Rare Disease Forum of Sri Lanka

The Sri Lanka College of Paediatricians co-organized the 6th Global Congress on Consensus in Child health in November 2017. At this congress an amazing variety of presentations by Sri Lankan pediatricians addressed a large number of rare diseases. Moreover, Sri Lankan pediatricians have recently established a Rare Disease Forum. To understand more about the care for children with rare diseases in Sri Lanka, a subsequent visit was planned. During this visit over four days, I was introduced to children in Galle, Ragama and Colombo, attended a meeting of the Rare Disease Forum and exchanged informal thoughts with colleagues.

Health system

Sri Lanka has a population of about 20.000.000 people. Healthcare expenditure per capita in Sri Lanka is about 200 dollars. To compare: the Netherlands has a population of 17.000.000 people, about 1000 pediatricians and a health expenditure per capita of over 6.000 dollars. The Sri Lankan health system is mixed public and private. Pediatricians serve in the public health system and have their private practices, separately. Almost all of the health data are recorded on paper. There are two separate files: the medical file which is held by the physician and a personal (note) book held by the patient or the family. The book is most complete and up-to-date. During each visit of the patient, the health care provider makes notes in the book including all the laboratory results. The patient/ family carries a separate folder with external laboratory test reports, x-rays, ultrasound scan reports, CT/MRI scans and other relevant reports belonging to the patient. When the patient has to visit more than one physician the number of books will grow however, every caring physician has access to all data related to the patient.

The pediatricians

Little over two hundred general pediatricians take care of the sick, chronically ill and disabled children in smaller (base) hospitals, middle large (general) hospitals and teaching hospitals. There are only a few pediatric subspecialists. Community pediatrics is an upcoming sub-specialty branching –off from pediatric training. After a five-year course for basic medical degree (MBBS) and one-year internship, pediatricians are trained for further five to seven years in a curriculum comparable to that of the UK. There is one year obligatory overseas training (either in UK, Australia or New Zealand). In one of the discussions, it appeared that Sri Lankan pediatric surgeons can do an European board exam.

In every teaching hospital visited, the Nelsons Textbook of Pediatrics was available at the pediatric ward. In addition, some departments possesses a copy of Smith Recognizable patterns of human malformation. At their work pediatricians have little or no access to internet, except through their personal mobile phones. To share clinical observations, pediatricians submit research papers and case reports to pediatric journal both locally and internationally. The editor of the local journal (Sri Lanka Journal of Child Health; open access link- https://sljch.sljol.info/) has over 2000 publications on rare diseases. There is a start with the introduction of electronic registrations in the hospitals however, mostly for hospital administrative purpose. Some pediatricians collect electronic data on their own.

Diagnosis and management of children

Most diagnosis of rare diseases in Sri Lanka are based on clinical observations and simple laboratory tests and /or images. For confirmation of a clinical diagnosis by DNA analysis, pediatricians are dependent on their relations with foreign DNA laboratories and on the budget of the parents. A national neonatal screening on hypothyroidism has just been introduced. During my visit, a broad

spectrum of children with rare conditions was presented. To be mentioned: Goldenhar syndrome, thalassemia (at center), Leri Weill syndrome, Spondylocoastal dysplasia, Glycogen storage disease, dyskeratosis congenita, metabolic disease, hyperinsulism, Prader Willy syndrome and hemihypertrophia. Most diagnosis are made on the basis of clinical observations.

Thalassemia is common in Sri Lanka and the level of healthcare is high at thalassemia centers. There is a close collaboration between the Thalassaemia center at Ragama with the Hematology department and the Weatherall Institute of Molecular Medicine of the University of Oxford in the UK. A few Sri Lankan physicians/pediatricians have been trained in Oxford and contributed to new insights in the management and treatment of children with thalassemia.

At the multidisciplinary clinic of the University of Kelaniya, Ragama, for children with disabilities, management is supervised by a pediatrician and speech- and physiotherapist perform early interventions. Children come directly from school for these programs. There are no special schools for children with learning problems. The clinic is familiar with the International (WHO) Classification of Functioning, Disability and Health (ICF).

The parents
Parents can visit the pediatricians whenever they wish. There is no appointment system, they come within a time span and wait their turn. The parents are well informed since they are the carriers of the data. Being a well literate country with female literacy rate over 95%, it seems that mothers are very capable of managing their child's health care and safe-guarding the child' s medical file. The pediatricians are often the only persons with whom parents, mothers, can share their personal concerns about the social impact of the child's condition. Most of the time these concerns have to do with the fear of exclusion and discrimination by society and reduced chances of getting a future marriage partners for their children. Chances of divorce, which is uncommon and socially un-acceptable in Sri Lanka, of couple with a disabled child is higher than average. When that happens the single parent and disabled child may even become more isolated. There is a fair amount of consanguineous marriages. So far there seems no need for parent groups except in the case of thalassaemia (link to patient support group- http://www.lankathalassaemiacircle.com/), or maybe the idea has not been introduced.

Medical costs
In Sri Lanka a chromosome laboratory is available. The cost of a karyotype is about 5000 LKR (27 €). The karyotype is not included in payment of public practice. Out of pocket payment is an obstacle for the lower class families. A onetime investigation would help to select the children with a chromosomal disorder to enroll in preventive management programs. As for example Down syndrome, with a high risk (40-50%) on a cardiac malformation. The price for a cardiac ultrasound (echocardiogram) recommended in Down syndrome is about 3000 LKR (16,50 €) and covered in public health. Early screening on cardiac malformations in children at rick improves quality of life and reduce costs by prevention unforeseen complications.

In conclusion
In Sri Lanka a relatively small group of well-trained pediatricians with a broad and large experience is capable of diagnosing and managing children with rare and disabling conditions. Since there are few patient organizations, at present pediatricians may be the best contact to reach the families and guard the child rights.

What could be initiated by collaborating European pediatricians and the Sri Lanka Rare Forum considering to support pediatricians and parents in their efforts to achieve the best possible health and wellbeing for the children with rare and disabling conditions

1) Establish a **minimal common dataset** including the presenting symptom(s) and time of diagnosis of rare diseases
2) **Pool pseudo anonymized** data to create large populations of the diagnosed children for epidemiology, natural history and research in Sri Lanka
3) Emphasize the need of access to **metabolic laboratory**
4) Emphasize the need of access to **chromosome investigations**
5) Raise awareness of rare diseases in the **pediatric training programs**
6) Support rare disease **patient groups**
7) Advocate for **humanitarian rights** of the disabled child on the highest attainable standards of physical and mental health.

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