

Teaching Family Medicine Approach in paediatrics with a patient having genetic syndrome – A case study

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I. INTRODUCTION

Family Medicine (FM) is an applied and integrative discipline. In 2011, the Ministry of Health introduced FM Residency Program in Singapore to train aspiring Family Physicians (FP). There are three sponsoring institutions, namely, Sing-Health Polyclinics, National Healthcare Group Polyclinics, and National University Health System (NUHS).

The Singapore FM Residency programs have rotations involving 13 disciplines, one of which is Paediatrics. As part of training in Paediatrics, the NUHS program has set up an outpatient Paediatric Longitudinal Clinic (PLC) staffed by FM Residents. This PLC is a service cum training clinic. It is supervised jointly by a Paediatrician core-faculty and a FM core-faculty. The FM Residency program aims at training a FP to provide primary, personal, and preventive care; and comprehensive, continuing and coordinated care to the patient and their family (Goh & Ong, 2014).

We report a case study of using a patient with a genetic syndrome to illustrate the Family Medicine approach in the training of the FP in a FM Residency Program. This Approach consists of the 3 Ps (personal, primary, and preventive care) to define care needs; and 3Cs (comprehensive, continuing and coordinated care) in managing the patient and family.

II. OBJECTIVE AND METHODS

The objective of this paper is to illustrate the potential of using FM approach in managing a patient with a rare multi-system genetic disorder. This teaching event took place at the PLC. The patient was clerked by the FM resident on duty. The one-year-old patient who looked dysmorphic to us (having microcephaly, broad thumbs and great toes, poor growth, and mental slowness), presented with constipation. The resident discovered that he was regularly seeing a neonatologist, neurologist, dietitian and gastroenterologist for the multiple problems he was having.

Using the FM approach, we defined the patient's primary, personal, and preventive care needs and formulated a comprehensive, continuing, and coordinated care approach in managing the patient and family. Through this approach we hoped to reduce fragmentation of care, duplication of services, and forestall complications through preventive care.

III. LEARNING POTENTIAL FOR THE FM RESIDENT

All three subspecialists managing this patient have worked independently of one another. The neonatologist with the help of a dietitian was following up the patient for failure to thrive. The height and weight centiles were < 3% and this did not improve with nutritional intervention. The neurologist was following up for

microcephaly and gastroenterologist was treating the patient for constipation. In this particular instance, it was the constipation that brought the patient to Children's Emergency and she was followed up by us in the PLC a week later.

During the case discussion with the FM Resident, the General Paediatrician recognized that microcephaly, broad thumbs and great toes are prominent features of Rubinstein-Taybi syndrome which is a genetic syndrome. The FM faculty encouraged the resident to read about this condition and find out if the presentation of the patient matches with what is described in text

books as well as explore the strategies to reduce fragmentation of care and work with various members of the health care team to increase the integration of care (Newbrunner, Borthwick, Finn, Scales, & Pearson, 2017; Starfield, Shi, & Macinko, 2005). In the follow-up discussion, the patient's care needs of the 3Ps (primary care, personal care, and preventive care needs) were defined. With a better understanding of the genetic syndrome, the FM Resident was able to formulate the 3Cs of management care needs (comprehensive, continuing, and co-ordinated care) of the patient and family. Table 1 shows the scope of the 3Ps and 3Cs in this case study (Gandhi, 2016).

- Primary care – first contact care – treating the presenting symptoms symptomatically e.g. constipation; feeding problems.
- Personal care – the mother may raise questions of physical abnormalities seen in the child with genetic syndromes and these will need explanations.
- Preventive care – anticipation of constipation, feeding problems, growth abnormalities – will benefit the child and support the family in caring for the child.
- Comprehensive care – knowing the range of abnormalities that may occur in the genetic syndrome e.g. slow development, cardiac abnormalities will improve the total care of the patient and support the family.
- Continuing care – cardiovascular abnormalities may be accompanied by heart failure that need continuing care; similarly delay in development may also need continuing care by the child development specialist.
- Co-ordinated care – the family physician can be the coordinator of care by integrating the patient-centred family focused multi-disciplinary care.

Source: Gandhi, 2016

Table 1. Components of the family medicine approach in the clinical case of a patient with genetic syndrome

IV. DISCUSSION

The FM approach in the integration of care in Paediatrics is illustrated in this case study of a genetic syndrome. The potential of using such a clinical case is rich. What is also important is the paradigm shift from the focus of studying biomedical features of syndromic rarities to integrate care of the patient.

The Rubinstein-Taybi syndrome (Gandhi, 2016) encountered in this patient is one example of a genetic syndrome that can be used to teach the scope and breadth of the FM approach. There are other genetic syndromes that can be used, e.g., the patient with Trisomy 21; Duchenne progressive muscular atrophy, or Spinal muscular atrophy (SMA).

Some of the day-to-day medical problems found in such genetic syndromes can be dealt with by the Family Physician alone, e.g., constipation. Other problems may need shared care of two or more clinical disciplines, as for example, the care for mental retardation, skeletal abnormalities, organ abnormalities, and slow physical growth. These are common features encountered in genetic syndromes.

The 3Ps: Defining the primary, personal, and preventive care needs of the patient.

Primary care is first level care. Constipation is an outpatient problem that the FP can help.

Personal care is important in connection with monitoring growth and development and providing acute care for common problems. The FP needs to help the patient and family define the tasks and provide recommendations where these are needed.

Preventive care has its role too. The preventive treatment of constipation, orthopaedic problems, mental stimulation, can be jointly coordinated by the FP and the Paediatric subspecialists (neonatologist, neurologist, and gastroenterologist in this case study).

The 3Cs: Managing the comprehensive care, continuing care, and coordinated care needs of the patient and family.

Comprehensive care spans the physical domain of the patient's clinical features, social domain of the patient (parents, helpers) which will include the caregivers who

need to know the medical condition that the child is having, as well as symptoms and treatment that will be needed e.g. constipation, schooling, and other life-course preparation for the child. The treatment of this condition is based on symptoms.

Continuing care is needed. In view of the multisystem involvement, anticipatory care may be needed e.g., dental care, intellectual and social development needs of the child.

Co-ordination of care is also needed. The child is under the care of the neonatologist, neurologist, and gastroenterologist. The FP could certainly co-ordinate the care between the specialists and in this way reduce duplication of care.

V. MOVING FORWARD

The Family Medicine Approach in teaching the management of the patient with a genetic syndrome shifts the focus from learning just biomedical rarities to teaching a system of defining the care needs that can be remembered as 3Ps; and the management of these needs as the 3Cs.

Through the co-ordination of care, reduction in fragmentation and duplication of care can be achieved and the inter-professional collaborative practice helps to deliver high quality, safe patient-centred services to achieve best possible long term health outcomes. Through prevention, some of the complications of multi-system diseases can be reduced.

Notes on Contributors

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Declaration of Interest

There is no conflict of interest, including financial, consultant, institutional or otherwise for any of the authors involved in this manuscript.

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