罕見難治的代謝病

許鍾妮醫生

會否想過一些外貌有異、毛髮粗糙、智力受損的小孩，可能正受著代謝異常疾病(簡稱代謝病)之苦？威爾斯親王醫院兒科的許鍾妮醫生，剖析這些不幸的孩子及家庭所面對的困難——除了未必可以治愈，還可能因歧視而得不到應有的體諒。

許鍾妮醫生(右一)攝於病人活動
Dr. Joannie Hui (1st right) in an activity for the patients

「代謝病」是什麼？

代謝病的種類多達數千種，泛指身體的新陳代謝出了問題。例如較常見的代謝病——「黏多醣貯積病」，因身體的遺傳基因細胞缺少了能分解黏多醣的酵素，當細胞內的黏多醣無法新陳代謝時，便會在體內細胞堆積，損害各個器官。

這些怪病是否遺傳？

部分代謝病是自體發病，但很多時患者的父母親各帶一個造成疾病的隱性不正常基因；父母健康不受影響(即隱性基因攜帶者)，若孩子遺傳到兩個有病的隱性基因就會發病。子昕及兩位兄長(請參本刊第10頁)所患的「腎上腺腦白質缺乏營養症」情況有點不同，是X-性聯隱性遺傳；有病的基因來自母親的X染體，兒子有一半的機率會得病。

「代謝病」有何病徵？

有害物質的堆積是漸進過程，孩子在剛出生時並無異樣。隨著年齡增加，堆積在體內的物質可能損及病童的外貌、智能、內臟及骨骼關節，造成面容毛髮粗糙、腹部突出、關節變形、脊椎變形、手指屈曲僵硬、行動不便、視力或聽力障礙；甚至呼吸道會變窄，引起肺炎等併發症。由於每種代謝病的病徵不盡相同，醫生必須像偵探般仔細推敲，再經過複雜的檢查，方可確診。

病童得到什麼幫助？

這些罕見疾病的嚴重程度不同，但大多隨著年齡增加而逐漸惡化。部分患者可在發病早期進行骨髓移植，以減輕病情及延長壽命。部分病人可接受「靜脈注射酵素治療」以減輕病情；但由於費用高昂(每年約需數十萬元)，目前並不普及。威爾斯親王醫院的「代謝病門診」會為病人的親屬作遺傳諮詢，盡量減低同類病發生的機會。這些病人和家屬的經濟和心理負擔都非常沉重，極需要社會大眾的關心和支持；若希望關懷這些病童，可先從認識他們的需要開始。

香港黏多醣症暨罕有遺傳病互助小組
地址：九龍橫頭磡村宏禮樓地下「香港復康社區復康網絡」轉交
電話：2794 3010
網址：www.hk-mps.com
電郵：mps@hk-mps.com

香港中文大學兒科學系 威爾斯親王醫院包黃秀英兒童癌症中心

黏多醣症病童及家屬郊遊樂
Outing for the Mucopolysaccharideoses patients and family members

The Rare and Intractable Metabolic Disease

Dr. Joannie Hui

Has it ever occurred to you that some mentally retarded children with abnormal facial features and coarse hair could be suffering from a metabolic disease? Dr. Joannie Hui of the Pediatric Department of the Prince of Wales Hospital tells us about the difficulties faced by patients suffering from these diseases and also the stress encountered by their parents. The disease itself can be life-threatening. What makes it even more unbearable is that some of these patients may be subjected to discrimination by the public.

What Is Metabolic Disease?

Metabolic disease refers to disorder in the body's metabolism. There are many types of metabolic diseases. For example, mucopolysaccharidosis, a common metabolic disease, occurs when there is a deficiency of specific enzymes needed to break down mucopolysaccharides (a kind of sugar molecules) in the genetic cells. When mucopolysaccharides cannot be metabolized and are stored up in the body damage will occur in various organs of the body.

Is the Disease Inherited?

Some of the metabolic disorders aren't genetic. But in most cases, the disease is caused by inheriting abnormal genes from both parents. While both parents remain normal, the child who receives one mutated gene from each parent will be affected.

The adrenoleukodystrophy (ALD) that has been affecting Tze Yan and his brothers (Please refer to P.10) is slightly different from the metabolic disease described here. ALD is an X-linked recessive inherited disorder. A male child born with the mutated gene carried on one of his mother's X chromosomes will get the disease.

What are the Symptoms of the Disease?

The affected child may look normal at birth because it takes time for the noxious substances to accumulate in the body. When the child gets older, the substances in the body may cause damage to the child's facial features, mental ability and the functioning of various organs and joints. This may result in rough hair, hernia, stiff joints, curvature of the spine and vision and hearing impairment. It may even cause complications such as the narrowing of the respiratory tract or pneumonia. Since the symptoms for each metabolic disease are different, doctors have to make an extremely careful search for clues from the patient's medical history, physical examination and together with a series of complicated investigations before confirming the actual diagnosis.

What Kind of Support do Patients Get?

These diseases could become very serious. In most instances, the condition of the patient deteriorates with age. Some of them can receive bone marrow transplantation at the early stage of the disease to reduce its damage and prolong their lives. Some may be treated by intravenous injections of the missing enzymes which are being manufactured artificially by the drug companies. This however is not a common treatment as it is only limited to a few diseases. Also it is a very expensive treatment costing up to a few hundred thousand dollars a year for each patient. In addition to diagnosis and treatment, the Joint Metabolic Clinic of the Prince of Wales Hospital also provides consultation on genetic issues to families with metabolic diseases with an aim to help reduce the risk of passing on the disease in future pregnancies through counselling and prenatal testing.

These patients and their families have to face serious financial and psychological challenges. They need your support and concern. If you want to show that you care, you can start with understanding their needs .

Hong Kong Mucopolysaccharideoses & Rare Genetic Diseases Mutual Aid Group
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