Understanding Rare Bone Disorders

認識罕有骨骼疾病

小而同
罕有骨骼疾病基金會
Little People of Hong Kong

The University of Hong Kong
Knowledge Exchange Project
Little People Care Alliance

香港大學知識交流計劃
「小個子，大作為」關愛行動

LITTLE BUT NOT LESS

小個子，大作為

Preface

By Prof John C Y Leong, SBS, OBE, JP
Chairman, Hospital Authority and Emeritus Professor, The University of Hong Kong

Skeletal Dysplasia is a heterogeneous group of bone abnormalities with more than four hundred conditions. However, these conditions are relatively rare in clinical practice, with the condition occurring in 1 to 3 of every ten thousand births. Its rarity means that most doctors, including orthopaedic surgeons, have relatively little experience in treating these conditions, and the general public often have bias when they see such patients.

In my 38 years of active clinical practice in the public healthcare system in Hong Kong, I have come across a reasonable number of such patients. While most of them have short stature and disproportioned limbs, the great majority have normal intelligence. Given the appropriate modifications of furniture at home, equipment at work place, and other public conveniences such as toilets and transportation, these patients can function normally.

Recent advances in diagnostic techniques, and treatment methods, have benefitted these patients tremendously. As an orthopaedic surgeon, the ability to lengthen limbs safely and successfully have enabled us to contribute significantly to improving form and function of these patients.

In this booklet, the "Little People Care Alliance" has done a good job to render awareness of these conditions to the public, highlighted the possibility of effective treatment, and given examples of highly successful achievements possible in the careers of such patients.

I congratulate the authors and editors of this very useful booklet.

序言

梁智仁教授SBS, OBE, JP
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「骨骼發育異常」泛指不同類型的骨骼生長不正常，共有四百多種疾病。然而，這些疾病在臨床上卻相當罕見，每一萬名出生嬰孩中約有一至三名嬰孩得病。由於罕有，大部分醫生，包括骨科外科醫生在內，對治療這些疾病的經驗較少，而公眾往往對這類病人存有偏見。

我在香港公眾醫療系統38年的臨床經驗中，曾經遇過一定數量的這類病人。他們大多身軀矮小，四肢不相稱，但絕大部分擁有正常智商。只要把家具、工作地點的設備及其他公共設施作出相應的改動，他們便能正常生活和工作。

近年醫學上在診斷技術及治療方法均有所進步，令這些病人獲益良多。作為骨科外科醫生，能夠安全及成功地把病人的四肢延長，表示我們對改善病人的外形和活動能力有很大幫助。

通過這本小冊子，「小個子，大作為」關愛行動能夠令公眾關注這類疾病，明白到有效治療的可能性，並把這類病人獲得非凡成就的真實故事傳揚開去，實在值得讚賞。

這是一本非常實用的小冊子，我衷心祝賀所有作者和編輯。

Preface

By Dr York Y N Chow, GBS, JP
Chairperson, Equal Opportunities Commission

On behalf of the Equal Opportunities Commission, it is my pleasure to contribute the preface to this knowledge-exchange booklet, "Little But Not Less: Understanding Rare Bone Disorders."

This booklet serves a dual-purpose. First, it provides useful information for families of people with rare bone diseases, including treatment options and recent relevant developments. This is both timely and necessary.

Secondly, this booklet can be a vital tool for raising public awareness and challenging prejudice about dwarfism and rare skeletal disorders. After all, discrimination often stems from a lack of understanding. Even though most people with short stature can lead independent lives and make meaningful contributions to society, just like others, they are often mischaracterised due to limited knowledge about their medical conditions.

I continue to be deeply inspired by the real-life stories of resilience and strength shared here by these patients and their families, who have fought for their right to a life of equality and dignity. In the following pages, you will learn how, with the right support, they have overcome numerous hurdles – medical, emotional, attitudinal, and social – in their everyday lives, including in education and employment. Indeed, this booklet provides a valuable platform to recognise their courage and determination, while also dispelling misconceptions about them.

By removing the barriers of misunderstanding and bias, we can enable people with dwarfism to equally participate in our society, which is to all our benefit. I commend the "Little People Care Alliance" members — Little People of Hong Kong, Li Ka Shing Faculty of Medicine and St John's College, HKU — for their joint efforts in putting together this much-needed informational resource. Such collaborative and multidisciplinary initiatives are indeed necessary if we are to foster a truly inclusive society where there are no barriers to equal opportunities.

序言

周一嶽醫生GBS, JP
平等機會委員會主席

《小個子，大作為：認識罕有骨骼疾病》是一本促進知識交流的小冊子，我很榮幸代表平等機會委員會為這本小冊子撰寫序言。

出版這本小冊子有兩大目的。首先，它為罕有骨骼疾病患者的家人提供一些實用資訊，包括在治療方法及最新醫學進展方面。這些資訊都是適時及必要的。

其次，這小冊子可以成為重要工具，提高公眾對侏儒症及罕有骨骼疾病的認識及關心，糾正一些偏見。畢竟，歧視往往源自誤解。雖然大多數身軀細小的人士都能獨立生活，與其他人一樣可以對社會作出有意義的貢獻，但社會上仍不少人對患者健康狀況一知半解，因而產生錯誤認知。

這類病人及家屬，為爭取應有權利過平等和具尊嚴的生活而努力付出，他們分享了一些充滿堅韌和毅力的真實故事，令我再次深受鼓舞。當你們繼續閱讀這小冊子，就會明白如果他們得到正確的支持，便可克服日常生活裡許多障礙，包括在醫療、情感、態度及社會上的種種困難，並可衝破教育和就業方面的難關。事實上，這小冊子提供一個寶貴平台，肯定他們的勇氣和意志力，並同時消除人們對他們的錯誤觀念。

只要消除了社會上的誤會和偏見，侏儒症患者便可以平等地融入社群，這其實對所有人都有益處。在這裡我要衷心表揚「小個子，大作為」關愛行動的成員，包括「小而同罕有骨骼疾病基金會」、香港大學李嘉誠醫學院及聖約翰學院，通過他們共同努力，才能把這些社會急需的資訊彙編成冊。要建構一個包容社會，讓所有人獲得平等機會，這些跨界別協力推行的計劃實在是十分必要的。

About "Little People Care Alliance"

The Little People Care Alliance is an initiative that began in 2014 under the Knowledge Exchange Program at The University of Hong Kong. Partnered with Little People of Hong Kong and St John's College, Little People Care Alliance aims to increase public awareness on the difficulties that are faced by Little People.

Despite the difference in their size to the average-sized person, Little People are fully capable of achieving just like everyone else. However, incorrect public perception about their abilities and discrimination may occur with respect to educational and work opportunities. Little People Care Alliance promotes equality and respect which can help create and solidify a positive attitude towards those who are different.

Schools and teachers play a major role in the development and nurturing of children's minds, yet many may not be readily equipped to effectively deal with the needs of Little People. Little People Care Alliance held an open forum, inviting schools and the general public to raise awareness, and promoting an inclusive and unbiased attitude towards Little People at school.

Together with St John's College, a High Table event was held with Little People as guest speakers. Members from LPHK, academia, as well as clinicians and university students were able to hear first-hand about the hurdles of the guest speakers and how they overcame these in school and the workplace to achieve their ambitions.

This booklet is a compilation of interviews, stories and articles that raises awareness on how the general public can help to alleviate some of the obstacles that Little People face every day.

Through these events, Little People Care Alliance hopes to promote a more positive and accepting attitude towards those with rare bone diseases. By facilitating a platform for discussion between affected families, the education sector, and policy makers, it is hoped that there will be more support for Little People.

關於「小個子，大作為」關愛行動

『「小個子，大作為」關愛行動』在2014年啟動，是香港大學的一項知識交流計劃。「關愛行動」集合了「小而同」罕有骨骼疾病基金會及香港大學聖約翰學院的力量，旨在促使公眾對罕有骨骼疾病患者多加關心。

侏儒症患者只是在身體上與常人不同，但他們與其他人一樣完全可取得各種成就。可惜大眾對此類疾病仍有誤解，對患者仍有歧視，這可能令他們失去平等的受教育及工作機會。「關愛行動」宣揚人人平等、彼此尊重的理念，有助社會大眾樹立正面對待與自己不同的人士的態度。

學校及教師在學童心理情智的發育和培養方面負有重要的職責，但現時很多學校仍未準備好照顧侏儒症學童的需求。「關愛行動」有見及此，曾舉辦一場公開論壇，廣邀學校代表及公眾出席，提倡建成平等共融的校園，鼓勵及接納患兒入學。

「關愛行動」與香港大學聖約翰學院合作，在學院的高桌晚宴上邀請侏儒症患者擔任演講嘉賓。嘉賓為小而同罕有骨骼疾病基金會會員、教授學者、臨床醫護人員、大學生親述自己如何在校園裡、職場上雖遭遇困難，卻克服障礙而實現自己的抱負。

「關愛行動」出版的這本特刊，匯集了多位患者及其家人的專訪及故事，希望有助大眾思考如何幫助罕有骨骼疾病患者，減輕他們每日面對的障礙。

「小而同」希望，藉助上述活動，可幫助公眾加深了解罕有骨骼疾病，並以正面及接納的態度對待患病者。「關愛行動」亦希望提供一個溝通平台有助患者家庭，教育機構、決策部門彼此討論，以便對患者增加支援。

Message from Professor Danny Chan

Assistant Dean of Faculty of Medicine, University of Hong Kong
"Little People Care Alliance" Project Coordinator

My passions in life are my family, dining and research. I am blessed with a loving family; enjoy my time as a chef and restaurateur; and the fortune of a job that started my academic career as a researcher in biomedicine.

Working at the Royal Children's Hospital in Melbourne has given me a different perspective to life. Seeing the children with abnormalities and their courage in dealing with deformities made me realize how lucky we are. My wish is to discover the cause of skeletal diseases, and to help those in need in our society. However limited my contribution may be, I believe it will make a difference.

Growing up in Australia, I am accustomed to a social system embracing those who need special care. Since coming to Hong Kong, I found the local system is in need of many helping hands for improvement. As an academic, I value opportunities to reach out to the community. With the dedication and professionalism of my colleagues, and the will and determination of the patient groups, I am "over the moon" to be a founding member of the "Little People of Hong Kong" (LPHK).

In a short period of 12 months, LPHK has achieved much in promoting its vision and mission, bringing awareness of public institutions to the need for Hong Kong to become a caring and loving society that embraces equal opportunity for all without prejudice.

This booklet is published as part of a knowledge exchange programme launched by"Little People Care Alliance". It aims to reach out to educators in Hong Kong for their support; help the younger generation understand the values of accepting everyone as equal and so lend a helping hand to those in need.

「小個子，大作為」關愛行動項目統籌

香港大學李嘉誠醫學院助理院長 陳振勝教授 寄語

家庭、美食和研究工作是我畢生熱愛的三件事。我有美滿的家庭；享受曾經是廚子及擁有自己餐廳的日子，亦慶幸開展了研究生物醫學的事業。

在澳洲墨爾本皇家兒童醫院工作時，我的人生觀點大有變化。看見那些患有骨骼疾病的小孩勇敢克服身體上的缺陷，我深感享有健全體格的人是多麼應該感恩。我希望找出骨骼疾病的病因，幫助社會上有需要的人。縱使個人力量微不足道，也深信這份微力可促使世界有所改變。

我在澳洲長大，向來認為社會接納並照顧傷健人士是理所當然的；來到香港以後，才發現本地的制度還需要大家攜手合力加以改善。我身為學者，十分珍視有機會以自己的專業知識服務有需要的社群。幸得人緣薈萃，遇到幾位專業人士有心奉獻，而許多病人和家屬也意志堅定，眾人決定為罕見骨骼疾病患者成立一個支援組織，我很樂意加入，就這樣參與創立了「小而同罕有骨骼疾病基金會」。

基金會成立以來，短短一年內已頗見成績：我們積極宣講基金會的宗旨和使命，令各大公立機構注意到：應促使香港成為一個有關愛之心的社會，　提倡人皆平等、無所歧視的理念。

此書的出版，源於香港大學的一個知識交流計劃，也即「小個子，大作為」關愛行動，旨在向大眾尤其是本港的教育工作者傳播知識並呼籲支持，幫助下一代認識到平等接納傷健人士乃關乎道義，並為他們伸出扶助之手。

Message from Ms Serene Chu
Chairperson of Little People of Hong Kong

The first year after Nathan's birth was filled with doubts, fear, anxieties and loneliness. Most of those around us had little knowledge of dwarfism (or achondroplasia), including the paediatricians and therapists in the public hospitals. Any attempt to find a local patient support group also ended in vain.

The events of September 2013 completely changed my life and the lives of many. Our family was invited by Prof Danny Chan, Dr Brian Chung and Dr Michael To to feature in a documentary for "Pearl Report", which portrayed the challenges faced by children with rare skeletal disorders. The programme pulled together a group of doctors, professors and volunteers who discussed the possibility of setting up a foundation to help these patients, especially children. Within a few months, we founded the Little People of Hong Kong (LPHK).

The collaboration between doctors, researchers and patient families proved to have a huge impact. Various projects and campaigns were initiated in 2014-15—patient families and doctors educated the public about dwarfism in radio shows and seminars; biologists, university students and patient families promoted awareness of rare skeletal disorders on Rare Disease Day. Not to mention this educational booklet contributed by all members of the "Little People Care Alliance", which is an initiative under the University of Hong Kong Knowledge Exchange Project.

What we have attained today are the concerted efforts of all the members of LPHK. They showed me courage under the horror of the shade, and hope beyond the wrath and tears. To close, I quote the last stanza of William Ernest Henley's "Invictus":

Invictus

It matters not how strait the gate,
How charged with punishments the scroll,
I am the master of my fate:
I am the captain of my soul.

William Ernest Henley

I dedicate this book to all unconquerable souls.

小而同罕有骨骼疾病基金會
會長朱凱欣女士寄語

弈元出生後的第一年，我一直陷於疑惑、恐懼、焦慮和孤單。身邊的人大多數對侏儒症（醫學術語「軟骨發育不全症」）了解甚少，即便公立醫院的醫生和治療師也是如此。我希望找到本地的病人支援組織，卻遍尋不獲。

2013年9月的一連串事件改變了我的生命，也改變了很多人的生命。我們一家獲陳振勝教授、鍾侃言醫生及杜啟峻醫生邀請，參與拍攝「明珠檔案」，內容關於患罕見骨骼疾病孩子面對的挑戰。這個節目讓一群醫生、教授和義工得以相聚，討論成立一個基金會來幫助這類病人，特別是兒童。短短幾個月內，我們創立了「小而同罕有骨骼疾病基金會」（簡稱「小而同」）。

醫生、研究員和病人家庭合作，果真發揮了很大的影響。2014-15年間，我們推動了幾項計劃和活動：病人家庭和醫生一起在電台做節目、辦講座，幫助大眾認識侏儒症；生物學家、大學生和病人家庭一起在「罕見疾病日」傳播罕見骨骼疾病的知識。當然，攜手出力的還有香港大學知識交流計劃下推動的「小個子，大作為 — 關愛行動」，以及「小而同」成員共同製作的這本知識性書籍。

我們走到今天，是「小而同」全體成員共同努力的成果。他們為大家展示了敢於面對黑暗的勇氣，以及超越了憤怒和悲傷的盼望。我在此引述威廉．亨利的《打不倒的勇者》末尾一節詩句：

> ***打不倒的勇者***
>
> *哪怕道路多狹窄，縱使磨難接踵來；*
> *我的命運，我主宰，我的心靈，我統率。*
>
> *亨利*

謹將此書獻給所有不屈不撓的心靈。

INTRODUCTION
引言

Skeletal dysplasia is a general term referring to the abnormality of bone growth. In many cases, skeletal dysplasia will result in extreme short stature which is commonly known as dwarfism. Due to their extreme short stature, people with dwarfism are referred to as "Little People" in many parts of the world.

In this book, we focus on the medical, emotional and social challenges Little People face from birth to adulthood, using real-life examples to show how they, with the support of family and society, overcome those challenges to live a fruitful life.

「骨骼發育異常」泛指骨骼生長不正常所致的一類疾病，往往導致患者的身材特別矮小，也即患上俗稱的「侏儒症」。由於身軀特別矮小，在很多國家中，這類患者經常被稱為「小個子」。

在本書裡，我們集中討論「小個子」從出生至成年所面對的種種挑戰，包括治療、情緒、社交等各方面的難題，並以真實的事例展示「小個子」怎樣獲得家庭及社會的支持而克服困難，活出豐盛的人生。

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Public misconception of dwarfism

Q1 : Are people with dwarfism less intelligent?

A1 : In general, no. It is extremely rare to have intellectual disability in conjunction with dwarfism; only a few very rare types of dwarfism have been correlated to any form of mental handicap.

Q2 : Do people with dwarfism exhibit strange behavior and mentality?

A2 : People with dwarfism do not exhibit strange behavior or mentality any more so than the average person. Due to physical limitations, people with rare skeletal disorders may do tasks a bit differently from the average-height person. Toddlers may need to climb higher to reach things, while adults may require the use of aids such as step ladders and stools. Nonetheless, people with dwarfism talk, act and think like ordinary people.

Q3 : Can people with dwarfism work normally?

A3 : YES, they certainly can! Many Little People work in various fields, some even serve as professionals such as lawyers, doctors and social workers. Given job opportunities, they can be valuable members of society.

Q4 : Is dwarfism inherited genetically from their parents?

A4 : Largely NO. Over 80% of all Little People have average-height parents and siblings. Most skeletal disorders are caused by spontaneous (unforeseen) mutations at conception, though a few are due to recessive inheritance (meaning that 2 mutations of the same gene are inherited from both parents who are each carrying only one mutation.) If one of the parents already has that disease, his or her mutation can be passed on to the children, affecting the latter's bones. This is dominant inheritance.

Q5 : Can these conditions be prevented?

A5 : These are rare mutations that happen by chance in the making of germ cells, and there are no preventative measures against this.

Q6 : Is the mother able to discover whether or not her child has dwarfism while the fetus is still in the womb?

A6 : Generally, no, the mother is not able to detect the problem when she is pregnant, although there are some exceptions, including some fatal types of skeletal disorders. The majority of dwarfism cases are not detectable by ultrasound, even in the 2nd trimester of pregnancy. Invasive testing is available for some conditions in the early stages of pregnancy, however, due to the rarity of these conditions, these are not usually available unless there is a family history of dwarfism.

Q7 : Can an improved diet, supplements (such as calcium tablets), or more physical exercise cure dwarfism?

A7 : Largely, no. Since it is a genetic condition, the above methods cannot cure dwarfism. In a few types of skeletal disorders, hormone injections may help. Advances in medicine and genetic therapy is hoped to one day provide a cure for these rare skeletal diseases.

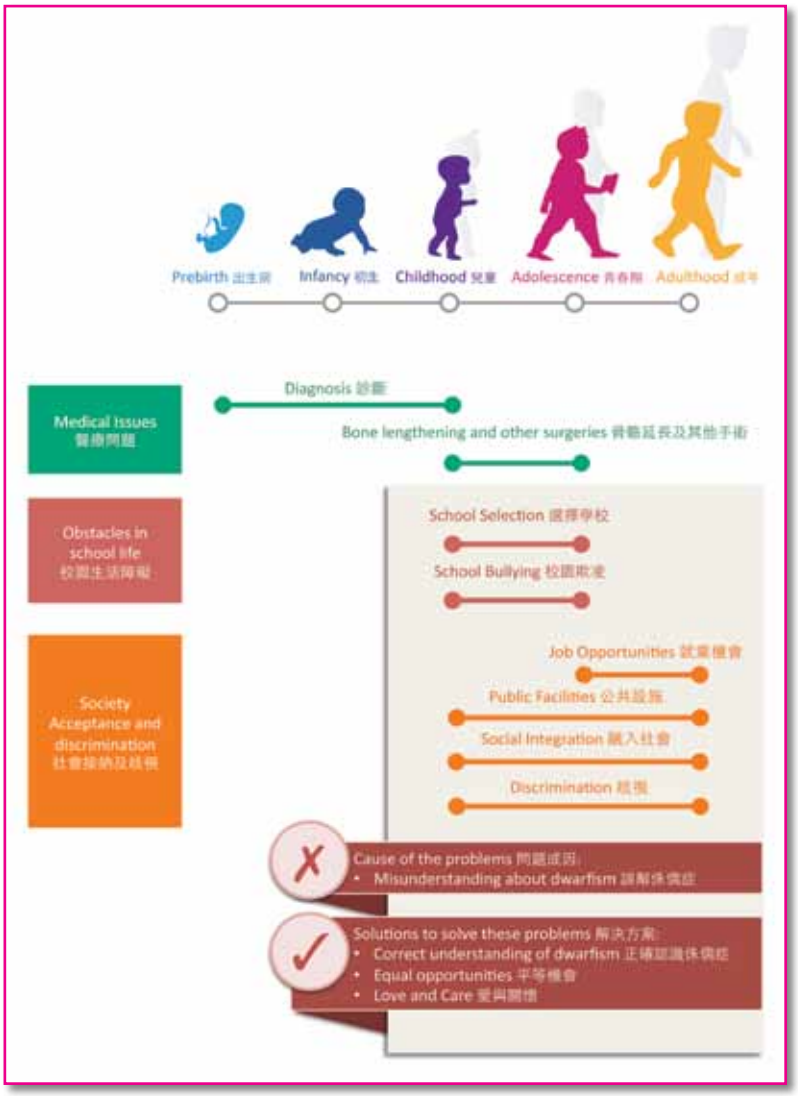
Q8 : Can the difference in height between a child with dwarfism and an average-sized child be narrowed as they get older?

A8 : No. The bone length, as well as the shape of bones of people with dwarfism are different from those of an average person, and also depends on the type of rare bone disorder they have. In general, the difference in height actually widens during adolescence.

Patients suffering from rare bone disorders, particularly those with dwarfism, seem to be subject to more teasing, or stares of astonishment and curiosity, than people suffering from the more well-understood disabilities including blindness, deafness, and wheel-chair-boundedness.

In both Asian and Western cultures, Little People have unfortunately been stereotyped according to their physical appearance. While sometimes Little People may encounter stares of curiosity due to their physical differences from the 'average person', sadly, they can also encounter inappropriate, rude and upsetting remarks from total strangers. In today's day and age, it is saddening to think that there are people who still prejudice against those who do not physically look like 'the norm'.

Seeking employment is a challenging task for any person, yet for a Little Person, it is a much greater struggle due to public misconception of their abilities. In the past, it was not uncommon for Little People to be hired as circus performers, or other employment in the entertainment industry such as actors portraying dwarves and other mythical creatures in movies and television. Other forms of entertainment such as 'dwarf tossing' were also used for amusement in bars, but thankfully due to changing times, this offensive and exploitative activity is now banned in a number of places.



We believe in the promotion of equality and social harmony, and hope to raise public awareness of rare bone disorders in order to eradicate prejudice against Little People.

關於侏儒症：大眾的誤解

疑問1：侏儒＝豬愚？患者的智商會不會較低?

答1　：一般來說，不會。患上罕有骨骼疾病而致身形矮小的人（侏儒症患者），一般都是智力正常；僅有極罕見的幾種侏儒症，才會與智力障礙有關聯。

疑問2：患有侏儒症的人，是否行為怪癖、心理異常？

答2　：當然不是！由於體格的局限，例如身體重心點偏低，侏儒症患者完成某些動作的方式或會不同，例如幼童會攀爬到較高處取物，成人則須藉助踏腳梯、矮凳等輔助工具，但他們的說話、行為、思想都和一般人無異。

疑問3：他們能正常工作嗎？

答3　：當然能。許多小個子從事各行各業，有的更成為律師、醫生等專業人士。只要有就業機會，他們都是能為社會貢獻一分力的。

疑問4：他們的病症是遺傳的嗎？

答4　：大部分不是。小個子之中，80％以上患者的父母及兄弟姐妹身高屬於正常。大多數骨骼疾病起因於受孕期間的自發性（非預料）基因突變，雖然也有少數病例起因於遺傳。其中有的是隱性遺傳，即父、母各自在同一個基因上含有突變但父母本人不發病，然而子女遺傳了父母的兩個含突變的同位基因而發病；如果父母之中有一方已是該病患者，其突變基因有可能以顯性遺傳方式傳遞給下一代。

疑問5：這些情況能不能預防？

答5　：這些屬罕見基因突變，在細胞形成的過程中隨機發生，沒有任何措施可以預防。

疑問6：胎兒仍在子宮內時，孕婦能否發現新生兒將會患上侏儒症？

答6　：一般來說，母親在懷孕時不能發現；雖然有一些例外，包括某些致命的骨骼異常。大多數侏儒症病例無法藉助超聲波檢查出來，即使早在懷孕第四至六個月時期做超聲波檢查，也不能發現。對某些病例，在妊娠初期可使用入侵式檢測方法，但此類病例極為罕見，除非針對有侏儒症家族史的孕婦，通常不使用入侵式檢測方法。

疑問7：改善膳食、服用食物補充劑（例如鈣片）、多做運動，能否治癒侏儒症？

答7　：大體說來，不能治癒。侏儒症是基因疾病，上述措施都不能治癒侏儒症。對少數幾類骨骼疾病，注射激素也許有幫助；當然，藥物的改良和基因療法的進步，有朝一日可望治癒這幾類的罕見骨骼疾病。

疑問8：患病兒童與正常兒童之間的身高差距，會不會隨年齡增大而收窄？

答8　：不會。侏儒症患者的骨骼長度及形狀與普通人不同，而且也會因罕見骨骼疾病的種類而異。一般而言，身高差距的擴大，實際上發生在青春期。

相對於患有失明、失聰等較常見的殘障，或行動不便而必須以輪椅代步的人士，罕見骨骼疾病（尤其是侏儒症）的患者似乎較容易受人譏笑，或引來較多驚訝好奇的目光。

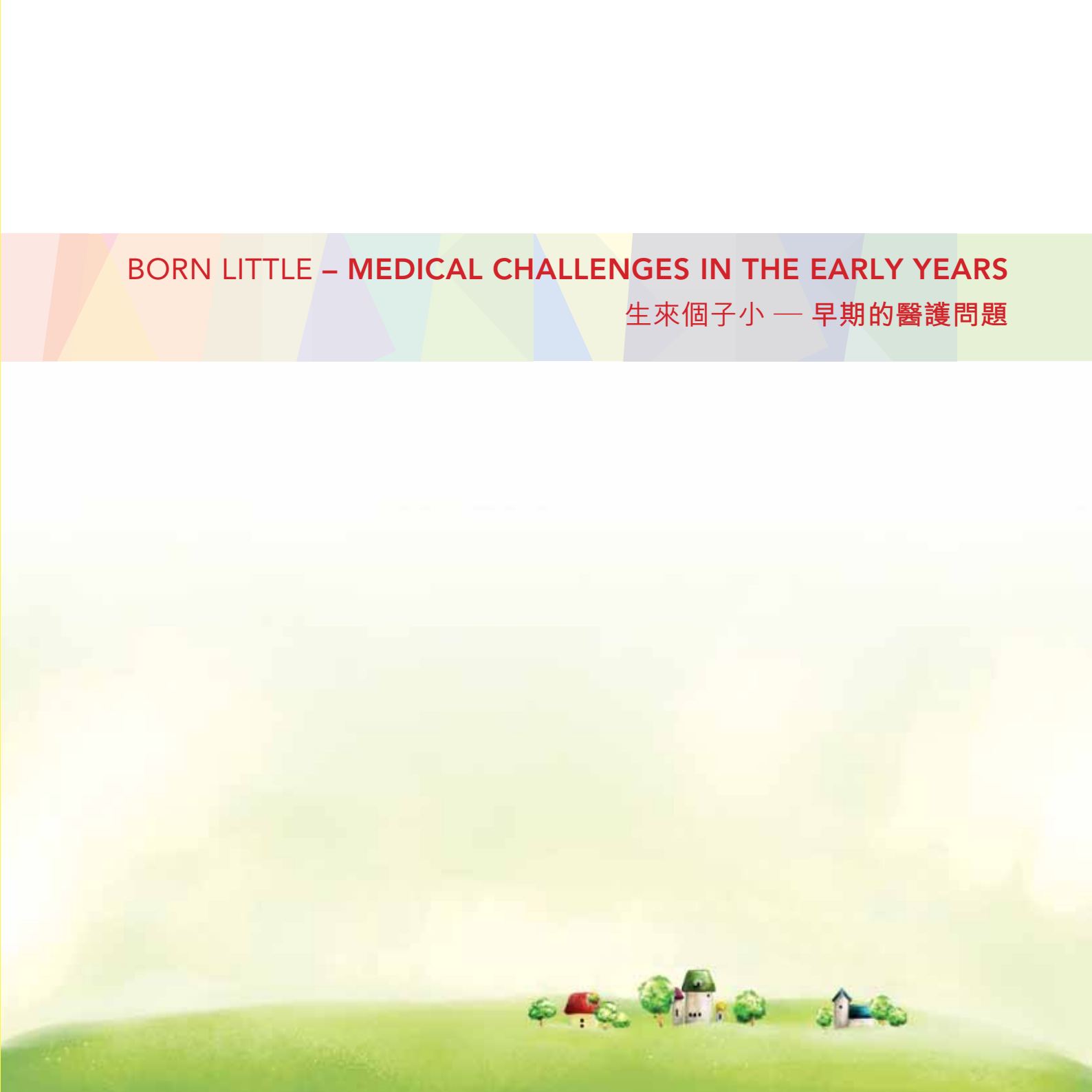
在亞洲和西方文化中，「小個子」往往因為身形被渲染而造成刻板印象。他們會因外表有別於「常態」而被投以奇異的眼光，亦可能受到陌生人不必要及不禮貌的待遇。在今時今日的社會中，仍然有人因為對方外表不同而作出歧視的行為，實在令人感到悲哀。

尋找工作對於一般人來說可能是一頂挑戰，但由於大眾對疾病的誤解，罕見骨骼疾病患者的就業問題更加嚴重。很久以前，「小個子」往往受聘於馬戲團扮演小丑，「侏儒」也常見於科幻電影及電視節目中扮演「小精靈」等另類角色。「拋小人」 亦曾經是在外國酒吧的消遣活動。幸好隨著時代的改變，這類嚴重違反人道的活動已經在世界各地遭到禁止。

我們希望透過宣揚平等及和諧社會，以及加深公眾對罕有骨骼疾病的認識，以期消除對「小個子」的歧視。

BORN LITTLE – MEDICAL CHALLENGES IN THE EARLY YEARS

生來個子小 — 早期的醫護問題



What is skeletal dysplasia?

Background information

Most skeletal dysplasias affect bone growth, and are often associated with dwarfism. It is a heterogeneous group with more than 400 conditions affecting human skeletal development. It occurs in 1-3 of every 10,000 births, thus making it a relatively rare group of disorders. In addition to short stature, patients with skeletal dysplasia may also encounter other medical complications, such as scoliosis (spinal deviation), osteoarthritis (degenerative joints) and neurological manifestations, depending on the specific cause of skeletal dysplasia.

Cause

Most types of dwarfism are caused by a spontaneous genetic mutation in the egg or sperm cell prior to conception. What prompts the gene to mutate is not yet understood although most are due to errors in DNA replication/synthesis during meiosis. The change is random and unpreventable, and can occur in any pregnancy. The following table shows the commonest causes of dwarfism and its occurrence.

Cause of disease	Occurrence
Achondroplasia	1/26,000 – 40,000 births
Spondyloepiphyseal dysplasia congenita	1/95,000 births
Diastrophic dysplasia	1/110,000 births
Growth hormone deficiency	1/3,800

甚麼是骨骼發育異常?

背景知識

「骨骼發育異常」泛指多種骨骼生長不正常所致的疾病。由於很多此類患者的四肢短小，身材亦偏矮，故此病俗稱「侏儒症」。有400多種發育異常的疾病會影響人體骨骼。每一萬個新生嬰兒中，大約有一至三人患此類病，因此是較罕見的疾病。患者不但身高比常人短小，也會有其他併發疾病，如脊柱側彎（脊柱向左側或右側彎曲）、骨性關節炎（關節退化）等，以及神經系統的問題，隨所患的骨骼發育異常之具體成因而異。

成因

多數種類的侏儒症起因於受孕前的卵子或精子細胞中自發的基因變異。基因變異的誘因尚不清楚，但大部分異變是來自減數分裂過程中的DNA複製/合成發生錯誤。如此變化是隨機發生、無從預防的，任何懷孕中都可能發生。下表顯示侏儒症的最常見病因及其發病率：

病因	病發率
軟骨發育不全	每26,000 — 40,000 新生兒之中有一例
先天性脊椎骨骨後發育不全	每95,000新生兒之中有一例
骨畸形發育不良	每110,000新生兒之中有一例
生長激素缺乏	每3,800新生兒之中有一例

What is achondroplasia?

Background information

Achondroplasia is the most common form of dwarfism with disproportionate limbs and trunk. The following tables summarize the possible clinical complications and their management by health care workers.

Clinical complication	Management
Hypotonia (low muscle tone)	Neurological examination
Obesity and risk of joint and cardiovascular problem	Growth documentation
Obstructive sleep apnea (paused breathing during sleep with or without snoring)	Sleep study
Middle ear dysfunction (affect hearing and risk of infection)	Auditory and speech evaluation CT/MRI of head and neck region
Spinal deformity and lower limb bowing	Orthopedic physical examination

Cause

80% of patients with achondroplasia have a new mutation (genetic change), with no affected parents. The remaining 20% are inherited from their parents in an autosomal dominant manner. The genetic mutation is located on the FGFR3 (fibroblast growth factor receptor 3) gene, which regulates bone growth and development. An individual with achondroplasia whose partner is unaffected has a 50% chance of conceiving a child with achondroplasia in each pregnancy. When both parents have achondroplasia, the child has a 25% chance of having normal stature, a 50% chance of having achondroplasia, and a 25% chance of having a lethal condition (i.e. having both copies of the abnormal FGFR3 gene).

甚麼是軟骨發育不全?

背景知識

軟骨發育不全是最常見的一類侏儒症，患者的四肢及軀幹比例異常。下表總結了可能的臨床併發症，以及醫護人員的檢查。

臨床併發症	臨床檢查
低肌肉張力	神經系統檢查
肥胖症、關節和心血管疾病的風險	身體生長記錄
阻塞性睡眠窒息症（睡眠中呼吸暫停，可伴有鼾聲，亦可無鼾聲）	睡眠分析
中耳功能障礙（影響聽力，並有感染風險）	聽覺及言語能力評估；頭部和頸部的電腦掃描或磁力共振掃描
脊柱畸形和下肢彎曲	骨科臨床檢查

成因

佔總數80%的軟骨發育不全症患者有基因突變，其父母並無發病。另外20%的患者則是從患病的父母遺傳而得病，遺傳方式為一體染色體顯性遺傳。基因突變的位置是在控制骨骼發育和成長的FGFR3（成纖維細胞生長因子受體3）。一個患有軟骨發育不全症的人與一個無此病症的人婚配，每一次懷孕中有50%的機率可產下患有軟骨發育不全症的嬰兒。如果父母雙方均是軟骨發育不全症患者，每一次懷孕中有25%的機率可產下身高正常的嬰兒、50%的機率可產下患有軟骨發育不全症的孩子，另有25%的機率可產下遺傳了雙重FGFR3基因變異的嬰兒，最後這類病症可致死亡。

Difficulties in diagnosis

Genetic disorders of the bone and cartilage occur through disturbances in the development, growth and homeostasis of the skeletal system. In the 2010 revision, the Nosology and Classification of Genetic Skeletal Disorders included 456 conditions of which 316 types are associated with genetic changes in 226 different genes. Commonly, these genetic disorders result in disproportionate short stature, a very common referral to paediatricians. Doctors have to detect all kinds of abnormalities in the size and shape of the limbs, trunk and/or skull of the patients through careful physical examination, radiographic evaluation and blood tests. Sometimes a biopsy of the bone and cartilage from the patient is needed to study the pathology. Since many genetic skeletal disorders are rare diseases of occurrence of < 1/2000 to 1/5000 of live births, it is very difficult for doctors to gain experience in these conditions. Referral to a clinical geneticist with expertise in skeletal dysplasia is often necessary to make the diagnosis.

Case study by Dr. Brian Chung:

Grace was aged 14 months when she first arrived at a clinical geneticist's clinic. With a body height of 66cm, which is below the 3rd centile, she was labelled with a diagnosis of Achondroplasia. However, genetic testing for Achondroplasia was negative. Upon detailed examination, Grace also had a prominent forehead, bifid uvula, flattened mid-face, and a small jaw. This was suggestive of type II collagenopathies. Bone X-rays further confirmed our suspicions and her genetic test of COL2A1 identified a recurrent heterozygous mutation, compatible with a diagnosis of the Strudwick variant of spondyloepimetaphyseal dysplasia. Genetic counselling was provided and her parents were happy that a diagnosis was finally reached.

The therapeutic value of a diagnosis cannot be emphasized more as it stops uncertainty. It helps carers and doctors to understand the needs of the patient and to stop an endless list of tests and medical appointments. It is also important to understand what it means for the rest of the family and to help the patient get services and support.

"The greatest gift to parents of a child with an orphan disease is to know a place where they can go and find a person who cares."

Dr J. Spranger (clinical geneticist)

診斷的困難

骨頭及軟骨的基因異常，表現於骨骼系統的發育、生長、體內平衡諸方面出現紊亂。2010年修訂版的《遺傳性骨骼疾病之分類》收錄了456種病，其中316種與226條不同基因的突變有關。這些遺傳性骨骼疾病通常令患兒的身形矮小、肢體比例異常，患兒通常會被轉介給兒科醫生診治。醫生需要用各種體格檢驗方法細心檢查患兒的四肢、軀幹、頭顱的形狀及大小，並使用X光評估及驗血等方法檢查。有時需要採集病人的骨頭及軟骨標本，作活體組織檢查，以供作病理分析。

很多遺傳性骨骼疾病屬於罕見病例，每二千至五千例成活的新生兒之中僅有一例， 所以一般醫生很難積累起經驗。此類病症必須轉介給專長處理罕見骨骼疾病的臨床遺傳學家，才能得出正確診斷。

鍾侃言醫生的案例：

Grace十四個月大時初次來到臨床遺傳學家的診所。當時她身高66cm，低於第3百份位數，她當時被診斷為軟骨發育不全症。可是基因檢測發現她並非患上軟骨發育不全症。經過詳細檢查，發現她另有一些表徵：前額凸出、懸雍垂（俗稱小舌）裂為兩瓣、面頰中部平坦、下顎細小。這似乎顯示有骨膠原二型病變。對骨骼作X光檢測後，進一步證實了我們的估測，而COL2A1基因檢測又發現一個復現性雜合基因變異，符合Strudwick型的脊柱骨骺發育不良的診斷。我們提供了基因學諮詢意見，她的父母終於得到一個診斷，感到滿意。

正確的診斷可消除疑慮，因而有利於治療，其價值無論如何強調都不過分。這有助醫護人員明白病人的需要，可免除一長串的檢驗及覆診。了解病人家屬的感受、幫助患者獲得診治服務及支援，也十分重要。

「對於有罕見疾病患兒的父母，最好的禮物是讓他們知道在何處可找到一個真正關心孩子的人。」

Dr J. Spranger (臨床遺傳學家)

Physical challenges from birth to childhood

Children suffering from skeletal dysplasia very commonly present with short stature. Some of them may develop spinal abnormalities and limb deformities. The children as well as their caretakers may face different physical and psychological challenges when the children grow up.

Achondroplasia is one of the most common types of skeletal dysplasia in which the children develop short stature. The average adult height is about 4 feet. Their trunk height is relatively less affected comparing with the limbs making their short stature relatively disproportional. The children may also have an enlarged head due to hydrocephalus as a result of too much fluid accumulated in the brain, and hump at the back due to spinal deformities at the thoracolumbar region.

The gross motor development of children with achondroplasia may be slower than normal children. A delay of 3 to 6 months in achieving motor milestones is not uncommon in most infants with achondroplasia. However, conditions like hydrocephalus and spinal abnormality should be excluded if there is a progressive deterioration in their motor development.

Bowlegs are common in these children. However, apart from the eye-catching appearances, these children do not usually have many complaints. Braces are sometimes used in keeping the legs straight but they are usually uncomfortable and difficult to wear especially if the children are young with short and chubby legs.

The problems related to short stature might not be so obvious when the children are younger as the caretakers will take care of most of the activities. Their body height when comparing with the classmates in pre-school may not differ by too much. However, they grow on average much slower than their classmates. As the children enter kindergarten, they may encounter problems such as when using the bathroom, or public transport because of their short stature. Their physical appearances may pose some psychological challenges. They may start questioning their physical appearance when comparing themselves to their classmates. The caretakers may need to provide psychological support and help them develop good self-esteem.

嬰兒期至幼童期的體格障礙

患有骨骼發育異常的兒童通常身材較為矮小。有部分患者會有脊柱異常及四肢畸形。在成長過程中，患兒及照顧的家人可能面對身心方面的各種難題。

軟骨發育不全症是十分常見的一類骨骼發育異常，患兒身材矮小，即使成年後，一般也只有4英尺高。與四肢相比，軀幹的長度受疾病影響較小，使得全身各部分的比例異常。患兒的頭顱可能因腦部積水而變大；而脊柱在胸腰段可能畸形，致使形成駝背。

軟骨發育不全症患兒的大肌肉發育比正常兒童慢，其中多數人要發育到能完成某些動作的階段，所需時間可能須延遲三至六個月。但如果患兒動作發育的狀況日益惡化，就應當排除腦積水及脊柱畸形，而另找病因。

弓形腿在此類患兒中很常見，但除了外觀問題外，通常一般情況下患兒不會有太多病症。有時可使用矯形來維持腿部正直，但通常令人不適且難以穿戴，尤其是在患兒年幼、腿部較為粗短之時。

年幼時，患兒身材矮小所致的問題也許不嚴重，因為他們的活動大多有家人照顧，而在學前階段，他們的身高與其他兒童也許相差不大。可是，平均而言，他們的長高過程比同齡兒童來得慢。進入幼稚園後，他們可能因身材矮小而在使用洗手間及乘搭公共車輛等情況下遭遇困難。外觀異常可能造成心理障礙，使他們在自感與他人不同時，可能開始對自己的外表發生疑惑。家人或照顧者可能須在心理上給予支援，幫患兒建立自信心。

A fatal piece of bone

Aria underwent surgery on the foramen magnum near the cervical spine when she was 14 months old.

Apart from universal neonatal health checks and immunization, Aria received various forms of regular examinations for her skeletal dysplasia after birth, which included skeleton, cranial nerve, hearing and vision examination, sleep study and brain MRI. At one follow up clinic, Aria was discovered to have mild signs of hydrocephalus (an abnormal accumulation of cerebrospinal fluid in brain), and the doctor suggested her to undergo an MRI investigation.

It was later discovered that a piece of bone in Aria's cervical spine was in a position more prominent than the normal vertebrae. The abnormal bone growth impaired the circulation of cerebrospinal fluid between the brain and the spinal cord, causing the signs of hydrocephalus.

After evaluation, the doctor commented there was a high risk for spinal cord compression if the piece of bone grew with age, which might lead to paralysis. Any accidental fall or risky motions in daily activities could also increase the pressure against the cord. Thus surgery was recommended to remove the piece of bone in the neck as soon as possible.

Children have great recovery ability. Aria was discharged from the hospital the following day after surgery. She could already manage to get up from bed on her own on the third day when she felt bored of staying in bed! Upon post-operation examination, there were no remaining signs of hydrocephalus.

At the age of 3, Aria now has checkups less frequently with her satisfactory performance in examinations and is undertaking early-phase training (including muscle growth, physiotherapy and occupational therapy) every month.

生死攸關的一塊骨頭

Aria在只有14個月大的時候，接受頸椎近處的枕骨大口（頭蓋骨基部的大洞，脊髓由此通向頭部）部位的手術。

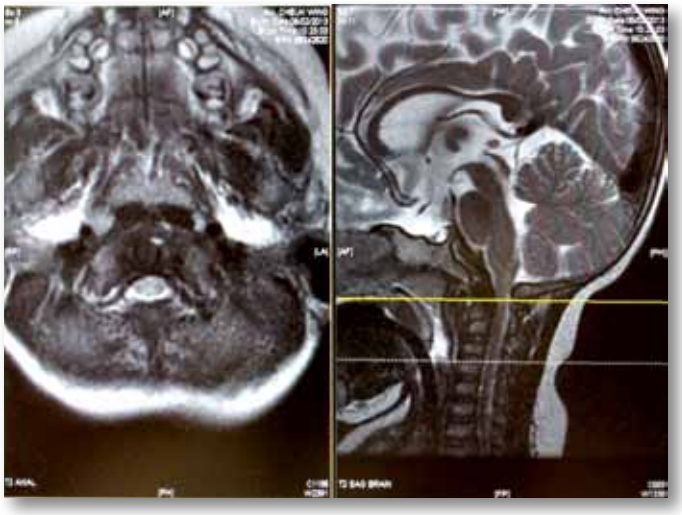
Aria患有骨骼發育不良症，從出生開始，除了接受一般嬰幼兒的例行檢查和免疫注射之外，還要接受多項定期檢查：骨骼、腦神經、聽力、視力、大小肌肉發育等檢查，以及睡眠測試和腦部磁力共振。在某次後續檢查中，醫生察覺到Aria有輕微的腦積水徵狀，於是建議她接受一次磁力共振掃描。

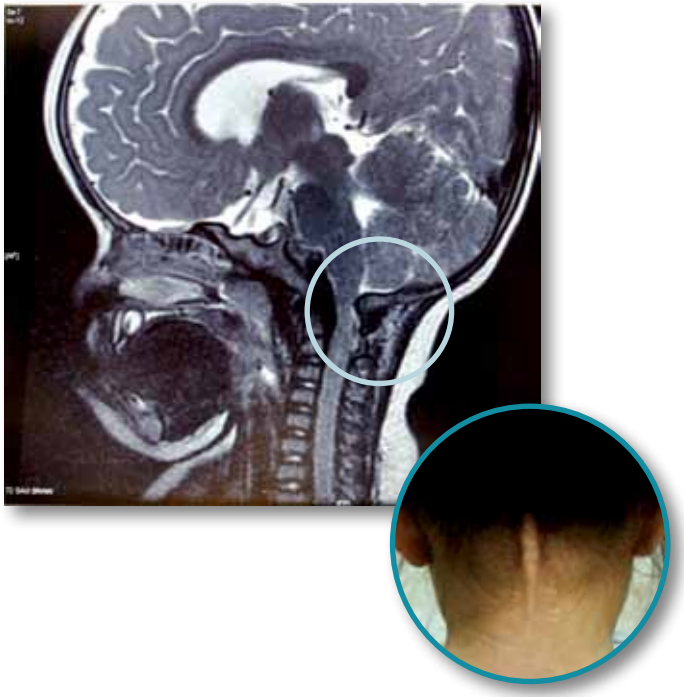
結果發現，Aria的脊椎部位有一塊骨頭的位置顯得異常，也即比正常的脊柱較為突出，這塊骨的生長阻礙了腦部與脊索之間的腦脊髓液的流動，造成了腦積水的徵狀。

經評估後，醫生說，那塊骨頭若隨年齡而增大，有很大風險會壓迫脊索而導致癱瘓，平日的不慎跌倒或危險動作，也可能增大骨頭對脊索的壓迫。因此醫生建議儘早接受手術，去除後頸處的那塊骨頭。

兒童的康復力旺盛，手術後第二天，醫生就讓Aria出院了。到了第三天，Aria已厭倦於繼續臥床，而能夠努力自行起床了！手術後複查時，發現腦積水的徵象已完全消失。

Aria現在三歲，由於複查結果令人滿意，如今已不需頻頻複查，目前正接受每月的早期訓練（包括肌肉發育、物理治療、職業治療等）。





The "little" angel who brings joy

Jayden was sent to the Neonatal Intensive Care Unit (NICU) immediately after birth at just 35 weeks gestation, weighing only 1.06kg with a body length of 34.5cm – falling below the 10th percentile of the average-sized baby of the same age.

The experienced gynecologist was at a loss to explain Jayden's slow growth rate during the pre-natal checkups, the differences in his facial features compared to the average baby, as well as his smaller-sized head, hands and legs. After staying in the NICU for two months, Jayden was finally allowed to go home after reaching the 4lb minimum weight required for hospital discharge. Since there were no similar cases at the Queen Mary Hospital, doctors could not make a diagnosis. Jayden's mother frantically searched online day and night for all the possible reasons that could be causing his symptoms, but could only narrow it down to diseases related to bone growth.

When Jayden was 9 months old, his trachea was found to be just 1mm wide and had the potential to suffocate him anytime. Doctors suggested performing a high-risk tracheostomy, however, his parents decided against surgery because they wanted Jayden to spend his life, albeit short, happily. Jayden had to therefore start living in hospital for his constant health check-ups. He was born a happy and sociable boy, often drawing people's attention with his laughter, and brightening up their day with his positive and optimistic character.

At the age of 1.5 years, Jayden was admitted to the Intensive Care Unit due to parainfluenza, with his condition so serious, he would die if he did not undergo surgery. This was the first time his parents openly wept at the hospital. After three long hours, Jayden came out of the operating theatre. The incision wound was so large in comparison to his tiny body size, yet Jayden's resilience was beyond anyone's imagination. Just two days after surgery, Jayden was already laughing happily as if there was no wound at all. His strength and perseverance touched the hearts of everyone around him, especially his parents, whom considered themselves so blessed have this little boy - an angel – who taught them how to love and treasure their loved ones.

Jayden was diagnosed with Majewski osteodysplastic primordial dwarfism type II (MOPD II) at the age of 2 years and 4 months - a serious and rare type of dwarfism, of which there are only approximately 50 patients globally. According to diagnostic predictions, this "little man" will have an adult height of around 100cm. While people may make frivolous remarks about his appearance, his heart is no different from that of other children. Jayden may not be physically fit, but given appropriate education and facilities, he has the potential to live a normal life.

Jayden's parents were especially glad to learn about the establishment of LPHK and their support of patients with dwarfism and other rare bone diseases and their families. They believe that through sharing experiences and supporting each other, all patients can have a positive attitude towards life. They hope that LPHK can raise public awareness of rare bone diseases, so that patients will no longer be embarrassed by the stares of strangers. LPHK's motto "We are little, but not less!" echo within their hearts.

帶來喜樂的「小」天使

Jayden一出生，就被送進新生兒深切治療部搶救，當時懷胎僅35週，體重只有1.06公斤，身長34.5厘米，這些數據低於同等懷胎期的新生兒之中第十個百份位數。

婦科醫生為母親做產前檢查時，已發現胎兒的生長緩慢，面部特徵異於一般胎兒，頭顱、手、腿都細小，醫生儘管經驗豐富，也茫然無從解釋。Jayden在深切治療部留醫兩個月後，體重達到出院的最低標準即4磅重，獲准回家。瑪麗醫院從前無類似的病例，醫生不能確定Jayden的病因。他的母親日夜上網，發狂般尋找各種可能導致類似病徵的原因，但只能將範圍縮小到與骨骼生長相關的問題。

Jayden九個月時，被發現氣管只有1毫米闊，隨時可能窒息。醫生建議接受高風險的氣管造口手術，但他父母決定不做手術，只想讓孩子快樂度過每一天，即使生命短暫。因此，Jayden住進醫院，無間斷接受健康監測。他天生就是開心果，很喜歡與人交往，時常發出笑聲引人注意，樂觀開朗的性格令人一天歡樂到晚。

Jayden一歲半時，因患上副流感，被送進深切治療部，當時病情危急，不接受手術就活不了。那是他父母第一次在醫院裡當眾哭泣。三個多小時後，Jayden從手術室出來，他的身體十分細小，相形之下手術切口顯得很大，但他的恢復力之強超乎眾人想像，手術後第二天，他已經笑得開心，彷彿身上全無傷口。Jayden的堅強毅力令身邊人人感動，父母尤其如此，因而深感蒙恩領福而有了Jayden這個小男孩　—　他猶如一個小天使，教父母懂得如何愛護、珍惜親人。

Jayden兩歲零四個月時，終於被確診為MOPD II型原生侏儒症，這是一種嚴重而罕有的侏儒症，全世界大約只有五十多個病例。據診斷推測，患者成年時將會是身高只有100厘米的「小個子」。雖然一般人看到Jayden的外形可能評頭品足，但他的內心跟其他小朋友無異。他的體能也許不足，但只要得到適當的教育和輔助設施，他也有能力過正常的生活。

Jayden的父母得知「小而同」成立，而且基金會支持侏儒症等罕有骨骼疾病的患者及家人，尤其感到開心。他們相信，大家交流經驗、互助支持，可使所有患者都以積極態度面對人生。他們希望「小而同」能幫助大眾加深認識罕見的骨骼疾病，令患者不再遭受陌生人的奇異目光而感到難堪。「小而同」的信念—「We are Little, but Not Less!」正為他們道出了心聲!

Early intervention for dwarfism

Depending on the severity of the case, children with rare skeletal conditions usually have altered physical development, some of which can adversely affect their health and daily life as they age. But early intervention by physiotherapists, occupational therapists and speech therapists can help families and teachers to provide a suitable environment and opportunities to maximize the developmental potential and quality of life of these children.

Physiotherapy

Little People are generally prone to musculoskeletal and spinal problems and exhibit slower gross motor development in the early years. With early intervention by physiotherapists from a few weeks of age, some of these conditions can be improved and potential problems avoided.

Early infant stage

Physiotherapists can suggest to parents preventative strategies such as teaching the baby how to control his head and trunk, sit up, and avoid postures that can potentially cause unnecessary pressure on the vulnerable body parts.

Late infant to early school age

Exercises (both typical and aquatic physiotherapies) that focus on strengthening the muscles of the arms, shoulders, shoulder blades, trunk, and those that maintain core strength with minimal pressure on the lower parts of the body are encouraged. Building up better muscle strength can enhance a Little Person's physical strength, lower the risks of bone and spinal deformity and alleviate fatigue in later years. Other stimulation exercises can also improve a child's balance due to disproportionate body parts.

Occupational therapy

With smaller hands and chubby fingers, Little People may have difficulty in acquiring fine motor skills. Furthermore, extreme short stature also hinders their ability to reach items placed at a higher levels. In some cases, mobility is an issue. Hence, an occupational therapist can identify and use physical adaptations and exercises which can enhance a Little Person's independence and autonomy in daily activities, such as self-care, personal hygiene and schooling.

Early infant stage

Occupational therapists can advise parents how to adjust car seats and position high chairs that are best suited for children with dwarfism.

Late infant to early school age

Occupational therapists can monitor a child's accuracy and speed of fine motor tasks such as writing, cutting, buttoning and zipping clothes, tying shoe laces, and suggest possible adjustments or equipment prescription to remove the obstacles to self-care and learning. They can also assess a child's mobility and customise a wheelchair if needed. Further, they offer advice to parents and teachers on how to modify the home and school environments.

Speech therapy

A few dwarf children are late talkers, possibly because of lower muscle tone, hearing impairment or restricted floor play due to back issues in the infant stage. Abnormalities in the jawbones, midface hypoplasia and incompetent lips may sometimes cause speech alterations or difficulties. In any case, speech therapists can run speech and pronunciation practice sessions to strengthen mouth muscles and promote earlier speech and language development before the sensitive period lapses.

Overall, ongoing monitoring is needed such that therapists can modify the programme in accordance with the child's progress.

早期介入治療

患上罕有骨骼疾病的兒童通常表現出發育異常，隨具體病況，異常的情形有輕有重，有些異常會隨年齡增長而危及健康，並影響日常生活。物理治療師、職業治療師、言語治療師等醫護人員的早期介入，可幫助患者家庭和老師營造適當的環境和機會，儘量改善患兒的發育狀況，並讓他們充分發展潛能。

物理治療

「小個子」通常較容易有骨骼肌肉和脊柱方面的毛病，在幼兒階段，則有大肌肉發育遲緩。物理治療師若在患兒出生幾星期後即提早介入幫助，部分的毛病可得到改善，將來可能出現的問題則可能避免。

幼兒期之前段

物理治療師可向父母建議一些預防措施，例如教嬰兒如何控制頭部和軀幹的活動、如何起身坐直，教他們如何避免一些不良姿勢，以免容易受傷的身體部位受到不必要的壓力。

幼兒期之後段至學童期之前段

物理治療師會建議做一些動作練習（有普通的物理治療，也有水中物理治療），重點是增強手臂、肩膀、肩胛骨、軀幹等處的肌肉，以及增強身體首要用力部位的肌肉，同時卻儘量不對下肢造成壓力。增強肌肉力量的練習可提高患兒的體能，並降低日後發生骨骼及脊柱畸形的風險，也可減輕疲倦。對於因肢體比例異常而平衡能力受損的患兒，另有一些刺激練習可改善其平衡能力。

職業治療

侏儒症患兒的雙手較小、手指較粗短，因此可能難以學會細巧的肌肉動作；此外，因為身材短小，從高處取物件的能力也會受阻礙。在某些情況下，行動不便也會造成問題。因此，職業治療師可辨別情況，教導患兒在體力上如何適應、如何練習，幫助「小個子」增強獨立自主的能力，以求能完成日常活動，例如生活自理、個人衛生、獨自上學等。

幼兒期之前段

職業治療師會指導父母如何調節汽車內的嬰兒座位，如何擺正嬰兒專用的高座椅的位置，以適合患兒使用。

幼兒期之後段至學童期之前段

職業治療師會追踪觀察患兒完成一些細巧動作（例如如寫字、剪東西、穿衣時的使用鈕扣及拉鍊、綁鞋帶等）是否準確、快速，並建議應如何調整動作、使用甚麼輔助設備，從而消除障礙，以助患兒生活自理，獨自上學。他們也會評估患兒的活動能力，必要時也會為患兒度身設計專用的輪椅。此外，他們可為父母和老師建議如何改建家居和學校的環境，以適應患兒的身體特點。

言語治療

少數的侏儒症患兒較遲學會說話，可能的原因包括肌肉張力較低、聽力有障礙，或因嬰兒期受背部毛病所限而較少在地上玩耍。在部分情況下，齶骨的發育異常、面頰中部發育不全、嘴唇活動能力不足等，也會造成發音不正或說話困難。無論原因為何，言語治療師都能指導患兒作言語練習、發音練習，以增強其口部肌肉，以便在幼兒的語言敏感期之內，及早促進其發音及說話能力的成長。

總而言之，各類的治療師都須持續觀察患兒的成長，以便因應其進步的情況而調整治療或訓練的計劃。

Current advances in treating dwarfism – limb lengthening

Limb lengthening is a procedure that helps to lengthen the bone in order to increase the body height. In general, limb lengthening can be performed in both the tibia and femur. Similarly, the upper limbs can also be lengthened after the lower limbs have undergone lengthening to improve the upper and lower limb ratio and to facilitate daily activities.

The technique of limb lengthening has been used for decades. The child undergoing limb lengthening will require general anaesthesia. The bone to be lengthened will be divided and stabilized by an external fixator, which needs to pass through the skin to anchor into the bone. The gap of the divided bone will be gradually widened at a rate of 1mm/day until the desired length is achieved. The lengthening process is not particularly painful but the children will experience tightness in the neighbouring joints as the length of the bone increases.

It is important that the children be compliant with the stretching exercise in order to avoid joint contracture. Depending on the amount of bone lengthening, the bone will require a few months to heal and restore the normal bone strength.

There are recent advances in limb lengthening using a nail that is inserted into the bone canal instead of using an external fixator. This new method will be more comfortable and more socially acceptable as the lengthening device is put inside the leg. However, the limbs of the children with achondroplasia are a bit short for the nails. Therefore, these intramedullary devices are not feasible for them at the moment.

Though many patients with skeletal dysplasia have short stature, not all patients with short stature will require limb lengthening. One must be aware that even if patients with achondroplasia undergo limb lengthening, they still cannot achieve normal body height. However, those who have received limb lengthening believe that the improvement in body height can facilitate their daily activities as well as self-image.

There is no consensus as to whether children with skeletal dysplasia and short stature should undergo limb-lengthening surgeries to improve their body height. Though they can have significant improvement in body height after limb-lengthening surgeries, the children and the caretakers must understand that the body height will still be shorter than normal. Limb lengthening surgeries can be usually done when the children become adolescents. Those who have undergone limb-lengthening surgeries, in general, are satisfied with the improvement in body height, self-image and their daily activities.

侏儒症治療法的新進展－上下肢骨延長手術

「骨骼延長手術」，顧名思義，就是藉伸長骨頭以增加侏儒症患者的身高。一般説來，小腿骨及大腿骨都可以用手術增長。與此類似，下肢經手術而加長之後，上肢也可藉助手術而伸長，以改善上下肢的相對比例，便利日常活動。

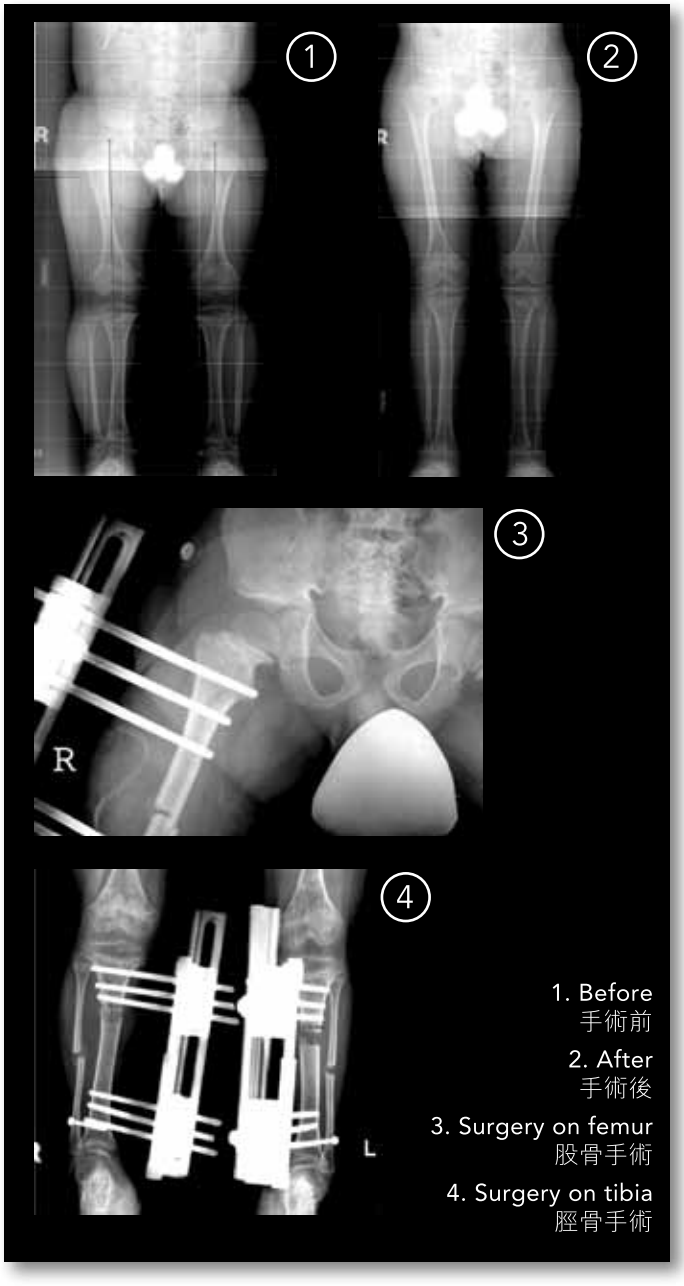
骨骼延長手術獲採用已有數十年歷史。接受手術的患兒需要全身麻醉。骨頭會被分割成兩段，然後將一枚外置固定器透過皮膚插入體內以鎖定骨頭。骨頭兩段之間的隙縫會以每天一毫米的速度逐漸增寬，直至整塊骨頭增至預期的長度。骨頭伸長的過程不會帶來太多痛楚，但在骨頭的長度增加時，患兒會感到相鄰的關節繃緊。

患者需要配合進行伸展練習，以防止關節攣縮變形。手術後需經過若干個月時間，骨胳才能癒合並恢復其強度，視乎骨頭伸長的程度而定。

近來的骨骼延長技術有了進步，可將一枚骨釘插入骨腔之內，以代替舊時的外置固定器。這種新技術使接受手術者較感舒適，且因為骨釘藏於腿骨內，大眾比較容易接受。可惜的是，與這種骨腔內藏型骨釘的長度相比，軟骨發育不全症的患兒的四肢仍顯得太短，所以目前這種療法暫不適用。

雖然很多骨骼發育不良的患者身形都很矮小，但並非人人都需要接受骨骼延長手術。須知軟骨發育不全症的患者即便接受過手術，其身高仍是不如正常人。可是，接受過骨骼延長手術的患者認為，改善身高有助便利其日常活動，也有助改善自我形像。

關於患有骨骼發育不良、身材偏矮的兒童應否接受骨骼延長手術以改善身高，目前仍無一致意見。雖然手術能顯著改善身高，但患兒及其父母要明白手術後的身高仍不及正常人。四肢骨骼延長手術通常可在患兒進入青春期時進行。接受過手術的患者，普遍對身高增加、自我形像改善、日常活動較為便利而感到滿意。



Current advances in treating dwarfism – drug development

Rare bone diseases are usually congenital, and the features can be observed at birth, with disability affecting individuals throughout their lives. Currently, over 350 unique disorders are described with a vast majority having no cure. Yet, studying rare bone diseases has helped elucidate biological pathways and revealed valuable targets for drug discovery that can benefit patients.

Traditionally, pharmaceutical companies are less interested in the development of drugs for rare disease patients as it is considered non-profitable. Fortunately, this mindset is changing, and industry is working with academic and research institutions for drug discovery strategies. It is clear that for success, we need to identify the knowledge gap in the biological mechanisms underlying rare bone disorders, and translate this to the bedside. As such, a multi-disciplinary approach comprising of researchers, clinicians, and patients is needed in order to advance our understanding of rare bone diseases, which can then allow for the development of novel therapies to improve patient outcomes.

There are now exciting examples of drug discoveries for rare bone diseases using state-of-the-art technology in precision medicine. One approach is to model human diseases in mice to gain a clear understanding of the disease mechanism. We can also take skin cells from patients and turn them into bone or cartilage cells in a laboratory, so that researchers can have millions of cells to screen for effective drugs. Drugs can be further refined and tested in mouse models, and with the help of pharmaceutical companies, these drugs may be brought to clinical trials in human patients. Indeed, this was an approach that was used to discover a potential new drug for patients with achondroplasia, a common form of dwarfism. The future will be a brighter one, and by working together, there is hope.

侏儒症治療法的新進展－藥物研發

罕有骨骼疾病通常是先天性疾病，病徵可在出生時發現，對患者造成的殘障則持續終生。現時有350多種獨特的罕有骨骼疾病，其中絕大部分沒有治療方法。然而針對罕有骨骼疾病的研究已有助於從生物學角度闡明其遺傳的路徑，也揭示出藥物探索的目標，新出的藥物可對患者有幫助。

照舊時慣例，製藥廠家為罕見疾病患者研發藥物的興趣不大，認為是無利可圖。可幸的是，如此心態正在改變，如今製藥業正在與大學及研究機構合作，以期發現新藥物。為求成功，顯然有必要先確認出罕見骨骼疾病的生物學機理方面的新知識，並將此知識應用於臨床診療。研究人員與臨床醫護人員的多學科合作，加上患者的配合，是加深認識此類疾病的必要條件，這才能促成治療方法的進步，以改善患者的病況。

目前，藉助精確醫藥學的尖端技術，罕見骨骼疾病的藥物研發已取得令人振奮的進展。研究的一個途徑是，利用小鼠模擬人類疾病以加深了解患病的機理；同時也採集患者的皮膚細胞，在實驗室裡將此轉化成大量骨骼或軟骨細胞，使研究者有數百萬個細胞可供篩選，藉以研發有效的新藥。此藥再經提純，並用模擬人類疾病的小鼠作測試；然後加上製藥廠家的協助，將新藥投入臨床試用，也即用於為患者治療。事實上，此法已用於研發出一種有潛力的新藥，將可用於醫治軟骨發育不全症，也即常見的一類侏儒症。可以相信，藥物研發的前景是光明的，只要各方群策群力，必將為罕見骨骼疾病患者帶來希望。

CHILDHOOD TO ADOLESCENCE – **LITTLE PEOPLE IN BIG SCHOOLS**

兒童期至青年期 — 小個子在大學校面對的挑戰

I'm Little: Have I a place in school?

Attempts by parents of children with rare skeletal disorders to gain a place into the school of their choice are fraught with many hurdles. Schools are often hesitant about the admission of these children as they know very little about their unique conditions and do not know how to care for them, physically and emotionally.

When Nathan, who has achondroplasia, was eight months old, his parents found a kindergarten which was willing to accept their son. Yet, three months before school started, they were requested by the kindergarten to pay an extra $7500 per month to hire an assistant for their son. "We were very disappointed because the extra school fees would be a heavy financial burden on the family and, most importantly, we did not believe our son required an extra full-time aide in school," said Serene, Nathan's mother. Fortunately, they eventually found another kindergarten which was willing to accept their son without extra charges.

The parents of Sam Lok, another preschooler with achondroplasia, also faced difficulties finding a mainstream kindergarten for their son two years ago. "During an interview with a principal, we were told that the kindergarten could not accept our son because she was worried that the other parents may not like to see a child with such a physical difference in school," said Win, Lok's mother. "We were very shocked and frustrated to hear such a statement," she expressed with a tinge of sadness.

Sadly, schools often have the misconception that having a child with dwarfism will create a manpower strain. In fact, with small adjustments and modifications, the school environment can be fully accessible.

我是小個子：有學校能接納我嗎？

患罕見骨骼疾病兒童的父母，為孩子爭取入讀所選學校的時候，往往遇到重重困難。學校不了解孩子的特殊情況，亦不懂如何在生理和心理上照顧他們，因此往往很猶豫，不願錄取這些孩子。

當患軟骨發育不全症的弈元還是八個月大時，他父母找到一間願意錄取他的幼稚園。然而，開學前的三個月，幼稚園要求他們每月多繳付七千五百元為兒子聘請一名助教。弈元的媽媽Serene回憶道：「我們感到很失望，因為額外的學費對我們家的財務是一大筆負擔，而最重要的是，我們不相信兒子需要一名全職助教在學校照顧他。」很幸運，他們最終找到另一所幼稚園願意錄取弈元，而又不索取額外費用。

另一個患有軟骨發育不全症的男孩岑洛，亦有相似遭遇。兩年前，他父母為他尋找主流的幼稚園時，遇到困難。他母親Win回憶道：「一次跟校長會面時，她說幼稚園不能錄取我們的兒子，因為擔心其他兒童的家長可能不喜歡看到學校裡有身體外形如此不同的孩子。我們聽到這種言論，感到震驚，也很洩氣。」說起往事，母親仍面露悲傷。

很不幸，學校往往有誤解，學生中若有侏儒症患兒，教職員工就要耗費精力去照顧。事實上，學校只要作出小小改建，校園設施就完全可供患兒進出並使用。

Facing challenges in schools

Little People are capable of attending mainstream schools, unless they have other learning disabilities that do not allow them to undertake the mainstream curriculum. Whichever school you choose for your child, be prepared to educate the staff about your child before school starts. Work with the school to create an environment in which your child can maintain independence and not be unduly physically stressed. It is a good idea to set an Individualised Education Plan with the school to help your child succeed in school.

Physical challenges in school

Little People may feel physically stressed in some sports activities such as long-distance running and hurdle events. Since Little People are more susceptible to neck trauma and joint dislocation, they should be under careful supervision when participating in certain kinds of sports such as gymnastics. If there is a need to change classrooms on different floors between lessons, Little People should be given sufficient time for travel or be allowed to use the lift. As junior patients tend to fall easily due to poor body balance, the school can educate students not to push Little People on the stairs to avoid danger.

Emotional challenges in school

Little People are often subject to curious stares or even bullying in school. Bullying can be in the form of teasing, mischievous jokes, physical abuse and isolation. To promote diversity and acceptance, the school can take initiatives to educate students, such as keeping a stock of books about dwarfism in the library and inviting adults with dwarfism to share their experience at assemblies. All school staff should have basic understanding on dwarfism and be able to answer simple questions from students.

身處校園，面對挑戰

患侏儒症的兒童都能入讀普通的學校，除非因有其他學習障礙而無法修讀主流課程。無論你為孩子選哪一類學校，要準備好在開學前就向學校說明孩子的情況。要跟學校合作，創造一個讓孩子可獨立活動而又不須過度消耗體力的環境。要幫助孩子在學校裡順利學習，最好與學校訂立一個孩子的「個人教育計劃」。

適應校園環境的困難

患侏儒症學生的體力可能不足以應付某些運動，例如長跑及跨欄等。由於他們的頸部較易受傷、關節較易脱臼，參加體操之類運動時，需有教師在旁細心指導。在兩節課之間，若需要轉換到不同樓層的課室時，應讓患兒學生有充足時間行走，或准許使用升降機。年紀小的患者平衡力較弱而容易摔倒，學校可教導學生不要在樓梯上推撞患者，免生意外。

在學校的情緒問題

患侏儒症的學生往往會遭人投來好奇目光，甚至被人欺凌。欺凌可有多種形式，可以是取笑、惡作劇、推打擠撞、排斥孤立等等。學校要鼓勵多元並存、接納差異的風氣，可採用多種方式教育學生，例如在圖書館提供有關侏儒症的書籍、邀請患有侏儒症的成年人在早會上講述自身經歷等。在學校裡，全體教職員都應有侏儒症的基本知識，並能簡單解答學生的疑問。

School modifications for Little People

General Building Access:

- Doorbell or handicap access button at a lowered height to open heavy exterior door
- Railing on stairs to decrease fall risk

Classroom:

- Supplies within reach
- Several stepstools for use by all children
- Heavy/bulky items modified or lowered
- Auto-on light switches, or lights left on until the end of the day
- Appropriately sized chairs. Physiotherapist or Occupational Therapist to help with fitting and adjustments
- Low stepstools or blocks placed under feet so that feet are not dangling
- Use floor chair if needed for back pain
- Low tables for floor-based work activities
- Monitor writing for fatigue – a soft splint may help later in the day or with homework for wrist instability

Playground

- High slides, monkey bars or other mega structures may be dangerous for LPs due to poor body balance. LPs should only use these facilities under close teacher supervision to avoid injury
- Due to increased risk of serious injury in case of severe head or neck trauma, avoid trampolines or playground equipment that can allow the child to hang upside down by the knees or feet

Bathroom:

- Preschool height toilet
- Stepstool for toilet and sink (skid resistant surface-wide platform preferred)
- Tape marks on floor to guide custodians and others for placement of stepstools
- Lower soap and paper towel dispensers
- Provide help if needed for personal hygiene. Private signal to teacher if require assistance to clean up after using toilet.
- Lower trash can
- Switch extender for lights, automated lights, or lights left on until the end of the day
- Cord extension from lever handle on both sides of door for access

為「小個子」改善校園設施

一般的無障礙大樓

- 增設門鈴或殘障人士按鈴（注意設置地點的高度），以供開啟較重的外門
- 在樓梯上增設扶手，以防跌倒

無障礙課室

- 應用的物品，須置於伸手可及之處
- 提供多個梯凳，供所有兒童使用
- 較重或較大件的物品，應設法改造，或避免置於高處
- 安裝自動燈光開關，或預設一天結束後自動熄燈
- 座椅的尺寸及高度應當合適；物理治療師或職業治療師協助安裝、調校
- 設置矮腳凳，以免坐者的腿腳懸空
- 為腰背疼痛者提供有軟墊的背靠椅
- 有地上活動時，提供矮桌
- 觀察患兒是否寫字時疲倦；對手腕肌肉疲弱者，提供軟夾板，以助其寫功課

遊樂場

- 侏儒症患者的平衡力較弱，玩高滑梯、猴架等有危險。小個子使用此類設施時，應有老師在旁照看，免生意外
- 兒童的頭頸部易受傷，因此應避免玩彈跳床，或須令身體倒掛的遊樂場設施

無障礙洗手間

- 設置學齡前兒童使用的較低的坐廁馬桶
- 在廁隔間內、洗手盆旁，提供踏腳凳（最好是表面防滑、基部較闊者）
- 在地板上，標明踏腳凳應安放於何處，以提示管理員。
- 將梘液及廁紙架的安裝高度降低
- 需要時，為患兒提供個人衛生清理。若須在患兒如廁後協助清理，發出私密信息以通知老師
- 降低垃圾桶的高度
- 為燈光開關加裝延長線，或採用自動燈光開關，或預設一天結束自動熄燈
- 在門板的內外兩面，為門把手加上延長繩，以便開門、關門

Little People-friendly schools

Ms Victoria Bewsey, principal of ESF International Kindergarten (Tsing Yi), and Ms Chan Chui-ling, principal of St Mark's Primary School emphasised the importance of creating an inclusive and supportive learning environment for children with special needs.

"We support children with dwarfism by setting an Individualised Education Plan and working closely with parents and support agencies. We teach children that 'being different is being special!' and there is no such word as 'Can't' in our kindergarten," said Ms Bewsey. The school tries to implement the curriculum with careful design to ensure all children can have full access of activities despite their physical difference.

The motto of Sheng Kung Hui St Mark's Primary School is "Never give up on anyone". "There is trust between the school, parents and the child. We believe in the students' abilities even if s/he has physical disabilities. They participate in sports day, field trips and other activities like everyone else," Ms Chan Chui-ling shared. To help new students and those in need to adapt to the school environment, they also have a programme called "Caring Angels", where older students from other year levels will serve as mentors for the younger students, including those with special needs.

關愛「小個子」的學校

英基國際幼稚園（青衣）的校長Victoria Bewsey女士及聖公會聖馬可小學的校長陳翠玲女士均強調，為有特殊需要的兒童創造一個包容而互助的學習環境至為重要。她們都有照顧侏儒症學生的經驗。

Victoria Bewsey 女士說：「我們透過『個人教育計劃』、與家長及支援機構合作，一同幫助患有侏儒症的學生。我們教導孩子說：『我的不同，就是我獨特之處』，而我們幼稚園不說『不可能』這個詞。」我們的幼稚園推行精心設計的課程，以確保身體有特殊狀況的孩子都能夠參與各種活動。

聖公會聖馬可小學的信念是「一個都不能放棄」。陳翠玲校長介紹說：「我們的學校、家長、孩子之間，大家互相信任。即使學生有身體上的障礙，我們也信任他們的能力。他們跟其他學生一樣參與運動競賽日、外出考察等活動。」為幫助新學生及有特殊需要的學生適應校園環境，聖馬可小學設有一個「愛心天使」計劃，讓高年級的大哥哥姐姐幫助低年級學生，包括有特殊需要的同學。

Little People-friendly schools

Sandy's daughter, Janice, is currently studying primary six in a local mainstream school, which provides her daughter with numerous opportunities and assistance. This fosters the joyful school life Janice has at the school, and encourages her to reach out to other classmates and her teachers.

"We are very grateful that the school has been providing a holistic approach to physical and psychosocial needs of a student," said the mother. "I recalled when Janice was in primary one, the school initially considered offering a set of desks from kindergarten for Janice. However, being worried about the possibility of discrimination from other classmates, the school placed a footrest under Janice's desk instead of opting for the kindergarten desk."

During primary two, Janice was once verbally bullied by a classmate. The school instantly took the initiative to handle the bullying incident and educated the whole school during morning assembly about the importance of mutual respect and good manners. This highlighted the significance of education in eliminating discrimination at school, which would otherwise impede the children's desire to learn and study.

Sandy hopes that in the future, the school will make further arrangements to facilitate her daughter to further adapt to school life. For example, she suggested reducing the weight of schoolbags, lowering the height of facilities like wash basins and blackboards, and equipping each floor with a squat toilet.

Sandy believes that education is of utmost importance when it comes to people with dwarfism. Intellectually, they are just like any one of us, and therefore as long as the environment is barrier-free, they can perform as well as we do.

關愛「小個子」的學校

Sandy的女兒昉誼現時在本地一所主流學校就讀小六，該校給了昉誼許多機會和幫助，使她的學習生活充滿歡樂，還鼓勵她與同學們交往，與老師溝通。Sandy說：「她常常跟同學有說不完的話，還常常主動幫助老師。」

昉誼進入小學前，小學老師曾考慮預備一套幼稚園的桌椅給她用，但又擔心她因此被其他同學另眼看待，最後決定仍用小學的桌椅，改為在書桌下加一個踏腳凳給昉誼擱腳用。Sandy說：「當時我心裡非常感激，因為學校竟這樣關懷一個尚未入讀的學生，在身體、心理上都照顧到。」

昉誼在小二時，曾有一個同學走到她身旁，在她耳邊細聲說她是侏儒。學校得知後，即時予以重視，在早會時提出此事，藉以教育全體同學應彼此尊重、舉止有禮。

Sandy說：「這樣的品德教育，對每個小朋友都非常重要。有良好品德，能消除歧視，對患有侏儒症的同學才最重要。侏儒症兒童其實沒有學習上的困難，只是往往處在歧視和欺凌的環境下，有人被迫放棄學習，這才是這些孩子學識偏低的原因。」

Sandy希望學校能作出一些調整，幫助侏儒症學童適應校園生活。她說：「校內各處的建築設計，應做到全無障礙，例如洗手盆、課室黑板等各項設施，應比正常情況略低，方便他們使用；再如，每樓層均設有蹲廁。另外，可設法減輕書包的重量。」

Sandy認為，教育對侏儒症患者非常重要，因為他們的智力跟常人一樣，只要有個無障礙的學習環境，他們的表現也會和其他人一樣。

Surviving the teen years

In face of bullying – the power of forgiveness

Manda, currently working as a warehouse supervisor, is a positive young lady and is thankful for everything she has in her life. Despite having skeletal dysplasia, she believes that a positive attitude can bring happiness in life. She has a caring family, a loving partner, lots of good friends and a job which she can make use of her abilities.

Nevertheless, there was still an unfortunate incident that happened in form one when she started attending a new secondary school. At the time, two of her classmates teased and verbally abused her because of her short stature. Since it was the first time she had ever faced such a situation, she hid in a corner and burst into tears at school. Luckily, this was discovered by her teacher and as a result, the two bullies were disciplined.

To her dismay, the disciplinary action imposed by the teachers did not stop the two bullies, and in fact worsened the situation. One day when Manda was returning home from school, she discovered that the lobby of her apartment building had been graffitied with foul language and her full name. This vandalism alerted her family and school authorities, and the two bullies were confronted by the teachers and Manda's mother.

To everyone's surprise, Manda's mother did not demand any form of further punishment for the two bullies, as she believed that only through forgiveness could hatred be resolved. She wanted to offer the two bullies an opportunity, just as she wanted everyone else to offer her child opportunities when she faced challenges and discrimination. This inspired the two bullies as well as many others, and eventually put an end to the bullying.

This story highlights the emotional challenges and bullying that children with skeletal dysplasia may encounter at school. But more importantly, it also inspires how we can handle adversities in life with compassion and wisdom.

順利度過青春期

面對欺凌－寬恕的力量

Manda現任職倉庫主管，她性格樂觀，對生活中的一切都慶幸而感恩。儘管患有骨骼發育異常症，她認為只要積極面對就過得快樂。她有溫馨的家庭，有位疼愛她的伴侶和很多的好朋友，亦有一份讓她發揮所長的工作。

但是，她開始上中學時，在中一那年遭遇一件不幸的事件。當時，兩名同學因她身材矮小而嘲笑她，甚至侮辱她。那是她第一次遭遇如此情況，她躲在一個角落裡大哭起來。幸好老師發現了，結果那兩名同學受到紀律處分。

令她失望的是，處罰沒有令那兩個同學改正，反而使情況更糟。一天Manda放學回到家，發現所住公寓樓的大堂遭人塗鴉，寫上粗言穢語及自己的姓名。這一破壞及侮辱事件驚動了她的家人和學校，那兩名學生被叫來見老師和Manda的母親。

出乎眾人意料，Manda的母親認為唯有寬恕才能消除怨恨，不要求對兩名學生再作處罰。她希望給他們一次機會，猶如希望其他人都能給Manda機會，讓她能克服障礙、免遭歧視。這啟發了那兩名同學和許多人，終於令欺凌結束。

Manda的經歷，突顯了骨骼發育異常的兒童在學校可能遭遇的情緒壓力和欺凌。更重要的是，這也啟發大家如何以同情和智慧來應對生活中的逆境。

The brittle child with a diamond heart

The darkest hour before dawn

Wai Fung, currently a 19 year-old adolescent, was born with cervical spine and rib fractures and diagnosed with Osteogenesis Imperfecta (OI). Worse still, he was subsequently diagnosed with OI Type V, of which there are very few cases in the world, and further complicated his diagnosis and management of his condition. Wai Fung suffered from multiple fractures since birth as a result of his condition, but many of those were minor fractures that could not even be detected by state-of-the-art computer tomography (CT) scans, not to mention the older types of technology used when Wai Fung was still very young. Consequently, this made the management of Wai Fung's condition more challenging and built up mistrust between himself and those surrounding him.

At the age of eleven, Wai Fung's conditions deteriorated to the point where he was no longer able to walk. His mother, being the sole breadwinner in the family (his father left when he found out his second son was born with a congenital defect), thought that Wai Fung was being rebellious and refused to walk. This delayed medical consultation and treatment, which further aggravated Wai Fung's mistrust against others. He became cynical and anti-social, and refused to listen to his mother, teachers and even medical professionals. This led to Wai Fung's "darkest hour", during which he quit school for a year in Grade 6, and the relationship with his mother had hit rock bottom.

A change in mentality

When Wai Fung eventually returned to school, he refused to take examinations and mocked his classmates and teachers. This in turn caused his classmates and teachers to dislike him, which eventually developed into a vicious cycle. At that time, everyone thought Wai Fung's future had no hope. Nonetheless, dawn always comes after the darkest hour. No one could pinpoint when the change in mentality happened, but Dr Michael To, his paediatric orthopedic surgeon at Queen Mary Hospital and Duchess of Kent Childrens' Hospital, was one of the first to spot this change and improvement of Wai Fung's attitude.

One day during Dr To's ward round, he noticed Wai Fung was reading a medical anatomy book he borrowed from hospital library. To Dr To's surprise, Wai Fung asked him many questions about the book and inquired about any correlation with his condition. The questions were not directed to blame anyone as he previously did, but to find out more about his condition and to seek improvement.

At that moment, Dr To realized Wai Fung had grown up from a grumbling child into a youngster with a more mature mindset. During the lengthy stay in hospital, Wai Fung continued to explore the world of books, and transformed his negative emotions into a desire for knowledge. Through reading, Wai Fung gained insight that knowledge could make a significant impact on his life, which fuelled his special journey through life.

Change in fate

When Wai Fung returned to school, he began to get involved with school affairs, and was appointed as chairman of his house in Form 3. He helped organize school activities and promoted students' welfare. This also helped him to reach out for an interpersonal connection and trust between him, his teachers and classmates. With his endeavor and appreciation from his teachers, he was promoted to chairman of the student union, something that no one could have ever imagined back in his "darkest hour".

In addition to school affairs, Wai Fung began to plan for his future and career. He shared with us his vision of being in the business field when he grows up, and has invested much effort to study for public examinations in the hopes of securing a place in university for a business degree. This was a sharp contrast to his attitude and performance during his "darkest hour", where he would play tricks on his classmates and teachers, and even skip classes.

As for his family, Wai Fung and his mother's relationship has greatly improved with Wai Fung developing a more mature outlook on life, despite the occasional dispute. Dr To also remarked that Wai Fung has become more cooperative during his consultations, and in fact, his willingness to share his story in this booklet with all of you is solid proof that he is starting to take steps in opening his heart to others. While we may not know what the future holds for Wai Fung, we can be certain that his "darkest hour" has passed, and that we shall witness a brighter future for him!

Osteogenesis Imperfecta (OI), also known as brittle bone disease, is a rare congenital disorder. Due to changes in the COL1A1 or COL1A2 genes, patients with OI are born with defective connective tissues and are thus prone to fracture, compared to normal people who are only bruised if subjected to the same force.

OI has a prevalence of approximately 6-7 in 100,000 live births. There are many subtypes of OI which vary in terms of features and severity. The complications depend on the manifestation of the affected individual. Complications include: fractures with minimal or no trauma without other factors (e.g. abuse or other disorders of bone), bone deformity, brittle teeth, hearing loss, respiratory problems (most severe in individuals with OI type II), tinted sclera (sclera is the white area of the eye) and more.

The current treatment depends on medications, exercise and nutritional advice to protect the bones from becoming more fragile, and promoting muscle and bone strength. Some patients can undergo surgical intervention for strengthening long bones, and prevent and/or correct deformities.

「玻璃骨」少年，有堅強的心

黎明前的黑暗

偉鋒現年十九歲，出生時脊椎頸段及肋骨有斷裂，診斷為先天性成骨不全症，更糟的是，隨後又診斷該病症屬於第五型，同類病例在香港乃至全球都極為罕見，令診斷及護理都非常複雜。由於患上此病，偉鋒自出生起又經歷了多次骨折，但骨折之處細如髮絲，即使最先進的電腦掃描亦難以察覺，何況是當年的舊式技術。這使得治療護理更加困難，令偉鋒對身邊的人失去信任。

十一歲時，偉鋒的身體狀況轉差，變得無法行走。他母親獨力支撐家庭（父親得知兒子患此病後就離家而去），誤以為偉鋒進入反叛期而故意不肯走路，因此耽誤了帶他應約上醫院診治，使得偉鋒對人更加不信任。後來偉鋒時常猜疑別人、不與人來往，對母親、老師、醫護人員所說的話都拒不聽從。在小六時陷入「最黑暗時刻」，停學了約一年，跟母親的關係亦跌至谷底。

心態的轉變

偉鋒終於回校上學，但拒絕考試，並羞辱同學及老師，令同學和老師不喜歡他，形成惡性循環。那時候，每個人都認為偉鋒的前途已全無希望。然而，最黑暗時刻過後，黎明到來了。無人能確知他的心理轉變何時發生，但最先目睹他情況改善的其中有一人是杜啟峻醫生，他是瑪麗醫院及根德公爵夫人兒童醫院的主診骨科醫生。

一天，杜醫生巡房的時候，發現偉鋒正在閱讀一本從醫院圖書館借來的解剖學書。杜醫生更感驚訝的是，偉鋒向他問起書中所述與自身的病況有何關聯。他的提問不再如先前那般一味抱怨，而是積極了解自己病況，尋求改善的方法。

那刻，杜醫生看到偉鋒已不是個只知抱怨的孩子，他已成長為思想較成熟的年青人。長時間住院期間，偉鋒繼續從書本探索知識，將消極情緒轉化成對知識的渴求。偉鋒從閱讀中得到的知識，對生活發生重大影響，推動他走過獨特的人生之旅。

命運的改變

偉鋒回校後，積極參與校園生活，在中三時被推選為社長，協助舉辦各種活動，並為同學服務。這份職責幫助他增進了與老師和同學的聯絡和互信。由於他的努力和老師的賞識，他當選為學生會會長，這是在「最黑暗時刻」中無人能夠想像的巨變。

除了學校事務外，偉鋒開始籌劃自己的未來和事業。他透露自己的希望是將來能投身商界，為此已並積極地為中學文憑試作準備，期望能入讀大學，攻讀商科學位。今天他的態度和表現，跟早先「最黑暗時刻」作弄同學和老師相比，形成鮮明對比。

如今偉鋒與母親仍偶有爭執，但母子關係大大改善，對人生的態度變得成熟了。杜醫生亦說，偉鋒樂意為本書讀者講述自己的經歷，就是願意敞開心胸對待他人的最好證據。我們無從預見偉鋒的前程如何，但我們深信偉鋒的最黑暗時刻已經過去，我們將看到他的未來變得光明！

成骨不全症俗稱「玻璃骨」，是一種罕見的先天性遺傳疾病。由於COL1A1或COL1A2基因的變化，患者骨骼中膠原纖維的結構比常人脆弱，輕微碰撞對一般人只會造成瘀傷，若發生在「玻璃骨」患者身上，就會引起骨折。

成骨不全症的發病率為每十萬個新生嬰兒之中僅有六七例。此症有多個類型，其特點和嚴重程度都各不相同。同一類型的成骨不全症的併發症，也會因患者的具體情況而有併發症，包括：無其他原因（如虐待或其他骨骼疾病）的骨折（創傷極輕微，或無創傷）、骨骼畸形、牙齒脆弱、失聰、呼吸疾病（在第二型病人最為嚴重）、有色鞏膜（鞏膜為眼珠的一部分，正常為白色），等等。

現時的治療只能靠藥物治療，鼓勵患者多做肢體練習、控制飲食，以防止骨骼脆弱加劇；練習和飲食也同時可增強肌肉和骨骼。部分病人可接受手術增強長骨，預防或矯正畸形。

Little People, Big Hearts

Growing up means facing many challenges, which is a basic fact of life. It is even more so for children with short stature, but how can we help them confront the challenges they may be facing? As adults who care about them, whether they be parents, friends, teachers, or professionals, we often focus on the problems. We try to find solutions, to protect them from harm, and even fight for their rights at a social and legislative level.

These may be important, but we often miss the even more basic factor: the attitudes and emotions of us influence the children most of all. Importantly, maintaining a positive attitude is crucial for children facing their difficulties, and can help them more readily overcome their hurdles.

For children, being different often means it is harder for them to find people with whom they can feel connected to. Yet children who feel loved and cared for often find it easier to connect to other people.

We need to love and care for them, but also need to let them express love. They need our support, but they also need us to believe that they can find meaning in life and become valuable members of society like everyone else.

Therefore, parents of children with special needs should not forget to take good care of themselves, and try to maintain a positive attitude. For parents of Little People, the pressure and frustrations may have begun since or even long before the diagnosis. While worrying about the needs of their children, they often forget to maintain their physical and emotional health and needs. Having a network of family, friends and good teachers not only benefits the children, but also the parents.

Do not be afraid to seek help for support. It can take a lot of searching and discussion, as not every teacher and professional are understanding and have adequate knowledge. Use every interaction as an opportunity to enhance the awareness of someone around you. Organisations like LPHK can help the children and their families expand their networks, and serve as a valuable resource to educate society.

Sometimes you may feel lonely. But remember, you are not alone.

為「小個子」開啟心扉

成長歲月，必有障礙，這是人生所難免。對於罕有骨骼疾病的患兒，成長中的障礙更大，我們可以如何幫助他們呢？身為成年人者，無論是父母、朋友，或是老師、專業人員，往往著眼於病況問題，尋找方法保護他們免受傷害，甚至為他們在社會上、法律上爭取權益。

這些當然都很重要，但我們往往忽略了更重要的因素：成年人自己的行為及情緒，對兒童的影響最大。自身保持積極的態度，對困境中的患兒至關重要，有助他們跨過障礙。

對患病兒童來說，身體與眾不同，往往很難找到可以同心傾談的人。不過，兒童若自感受人關懷，則比較容易與他人聯絡溝通。

我們需要關心愛護他們，但也要讓他們懂得如何表達愛。他們需要支持，但更需要別人信任他們能發現人生的意義，並能同大家一樣，成為對社會有貢獻的成員。

因此，有特殊需求的兒童的父母切勿忘記，應先照顧好自己，保持積極樂觀的心態。罕有骨骼疾病患兒的父母，通常在子女確診之後，甚至早在此前，深感壓力和沮喪。他們會憂慮患兒有何需求，卻疏忽了自己的身心健康和需求。與朋友、家人、學校老師建立聯繫網絡，不但對兒童有益，也對父母有益。

不要害怕向人求助。老師和專業人員並非個個善解人意，也非人人有豐富知識，因此，向人求助可能要經過許多摸索及交談。成年人應珍惜這些互動的機會，讓身邊的人加深關注及了解自己的情況。參與「小而同罕有骨骼疾病基金會」之類的病人互助機構，可以幫助患兒及其父母拓寬交往，亦有可能為大眾傳遞罕見骨骼疾病的知識。

有時候，你感到孤單；但請勿忘記，世上總有同路人。

A LONG AND WINDING ROAD FOR DIGNITY

爭取尊嚴的漫漫長路

Integrating with society

Due to their short stature, Little People find it challenging to reach things that are placed higher than 1.2 metres. Reaching the uppermost shelves in the supermarket or public library, even when on tiptoes, is almost impossible without the aid of a stool. Simply using a drinking fountain to get a sip of water, or using an automatic teller machine (ATM) are also unfeasible for Little People, as these facilities are often unreachable by Little People.

Another challenge for Little People is access to public toilet facilities. While the average person may not give a second thought about using a public toilet, the majority of public toilets are too high for Little People to use. This is in addition to the challenges of reaching the door handles and locks, the sinks, and the soap and hand towel dispensers. A lady with dwarfism in her early 20s recalled the troubles with the public toilet facilities she encountered during her teens. "I enjoyed going out to watch movies with friends, but sometimes it would be embarrassing when I needed one of them to hold me up to reach the tap to wash my hands".

Provisions and modifications under the Design Manual – Barrier Free Access 2008, issued by the Building Authority, which are mainly for wheel-chair bound, and visual and hearing impaired are not usually suitable for Little People. For example, the toilets and washing basins for wheelchair-bound people are considered still too high for Little People. Furthermore, the manual only applies to new buildings and existing buildings undergoing renovations, while many older buildings do not have lift control buttons at a lower level that are suitable for Little People, who often have to rely on the help of others.

On the other hand, in recent years, public transport facilities in Hong Kong have made modifications to buses and MTR stations which have greatly improved the accessibility of these facilities for Little People. The larger style buses are able to adjust their height to allow Little People to more readily board buses, and the MTR stations have made provisions of movable platforms and lifts with buttons at a reachable height. However, for other forms of transport such as the minibus, it is still difficult for Little People as they still need to overcome a big difference in height to step on board.

For the short-statured person, better thought-out design of facilities can greatly improve their dignity and independence.

融入社會

對於身材矮小的「小個子」，伸手取下置於高過1.2米之處的物件已是難事，若無踏腳凳之助，縱使踮著腳尖，要在超市或圖書館的高層架上取下書本、貨品，近乎全無可能。常見的飲水機和自動提款機，若是安裝位置較高，他們也很難使用。

「小個子」面對的另一個障礙，是使用公共廁所。大多數身高正常的人，使用坐廁馬桶可以不需思索，但這對患者來說又是一個難題。這跟開門、洗手、取用洗手液和抹手紙的情形一樣，因為這些設施都安裝在較高位置，他們手不能及，所以很難使用。一位二十來歲的患者回憶說：「當年很喜歡和朋友們結伴去看電影，但用了廁所後，往往要請朋友把我抱起來，才可以洗到手。」

建築事務署印發的《設計手冊：無阻礙通道2008》，主要的服務對象是輪椅使用者、視障及聽障人士，很多規定不適用於侏儒症患者。例如：傷殘人士專用的坐廁和洗手盆，為了照顧輪椅使用者，其位置都較高。此外，《手冊》只適用於新落成的樓宇，或是即將擴建或改裝的現有樓宇，而在較早時建造的樓宇內，升降機內的按鈕並非設在較低位置，患者可能需他人幫助才得以使用。

另一方面，一些公共交通的營運商如巴士公司、地鐵公司等，如今改善了巴士站、鐵路站等設施，例如大型巴士可以降低其活動登車平台，鐵路站的升降機內有安裝於較低處的按鈕等。這些改善，為罕見骨骼疾病改善了出入的便利。不過，乘搭小巴時，患者在登車時仍須克服地面與車身間的高度差。

對於身材矮小的患者，如果各類公共設施的設計能較多考慮他們的特別情形，將大有助他們增進尊嚴和獨立。

Job opportunities for Little People

"What we want is equal opportunity."

Patients with dwarfism often face tremendous difficulties and discrimination when seeking employment. Some employers hold a misconception that Little People have below-average intelligence. In fact, Little People only differ in stature from the average person, but not their intelligence or working abilities. Their performance can be even more outstanding as they cherish the precious opportunity more.

Social worker Edmond, who has achondroplasia, thinks that there are not many limitations in his choice of career although there are difficulties for him to take up specific task such as youth adventure training. He has devoted a lot of effort to prove that he is no different from his colleagues so that people have confidence in him. "I was lucky because my employer had no doubt in my abilities during the job interview. Yet, some employers do not like the physical appearance of Little People and think they will affect the corporate image," said Edmond.

Edmond believes there is very little support for job placements for Little People. He hopes the government or other organisations can provide training and job referrals for Little People who wish to join the work force.

According to the Selective Placement Division of the Labor Department, there were 9,369 job vacancies in 2012, including clerks, sales, customer service, laborers, and so on. Among the 2,686 registered job seekers, 385 are Little People and 299 of them are employed.

「小個子」的就業機會

「我們想要的，是平等的機會。」

侏儒症患者在求職時往往遭遇極大困難及歧視。有不少僱主對侏儒症人士存有誤解，認為他們的智力不如常人，但其實此類患者只是身材較一般人矮小，智力及工作能力卻與常人無異。他們往往比一般人更珍惜難得的機會，因此在工作中甚至表現得更為出色。

現職社工的Edmond患有軟骨發育不全症，他認為在選擇職業時沒有太大局限，但面對某些特定職責，例如青少年歷奇訓練等，就會感到難以承擔。他必須作出很大努力去證明自己與同事無異，以使人對自己有信心。他說：「我很幸運，僱主在面試時不懷疑我的能力，但另有些僱主不喜歡侏儒症患者的外表，認為會影響公司形象。」

Edmond認為，社會給予「小個子」的就業支援不足。他希望政府或其他團體能為有意進入職場的「小個子」提供培訓，並給予職業轉介。

據勞工署下轄的展能就業科的統計，2012年共有9,369個職位供人申請，包括文員、銷售、顧客服務、體力勞動等；有2,686人登記求職，包括385名侏儒症患者，而其中的299人獲得了聘用。

Little People who stand tall

A "little giant" on the badminton court

If you ever had the pleasure of meeting Tim, you would know in an instance that he is a very unique individual.

We are not talking about his physical appearance that makes him different from others, but the personality and spark that makes him stand out from others. An energetic and active young man who always has a heart-warming grin on his face, Tim works at an IT department of a financial company during the day – just like any one of us. But at night, he transforms into a superstar on the badminton court! Tim is a representative of the Hong Kong badminton team short stature group, and often represents Hong Kong in international badminton competitions.

In addition to sharpening his badminton skills and expanding his social network, participation in international competitions also broadens his horizons in the well-being and care for patients with rare diseases. "There is no doubt that there is greater support for patient groups in countries other than Hong Kong", Tim expressed when meeting other overseas sports players with skeletal dysplasia. He found that the scale of the overseas patient groups was larger than that of Hong Kong due to larger populations in other countries, which contributed to a larger pool of people with skeletal dysplasia. In addition, overseas patient groups had more support and interest groups to cater for a diverse number of activities ranging from sports teams for basketball, swimming and badminton, to study groups that assist with academic needs of younger patients, and other social activities such as hiking and dating.

When asked about LPHK, Tim agrees that "LPHK in Hong Kong is doing very well given that it is only at its beginning stage." He compares the local and overseas patient groups and believes there are many opportunities for LPHK to explore and expand on.

As for parents and educators, this encouraging story of Tim further highlights the importance of holistic care and education for people with skeletal dysplasia. "Support and love from family members is of paramount importance", as Tim recalls the trust his family and teachers had in him when he opted for a mainstream secondary school and eventually university.

小個子，大作為

羽毛球場上「小巨人」

如果你有幸認識Tim，一眼就可見此人不同尋常，這不是說他的外貌身型，而是他的個性和朝氣令他自成一格。這個活力充沛而好動的年青人，臉上常掛著笑容，令人感覺溫暖。他日間在某金融機構的資訊科技部門工作，晚上就一變而成為羽毛球場上的巨星。他是香港羽毛球代表隊（身型矮小組別）的成員，經常代表香港參加國際比賽。

Tim接觸了患罕見骨骼疾病的外國運動員後，感慨說道：「外國現時對患者的支援確實比香港多。」他看有些國家的人口龐大，患罕見骨骼疾病者的人數亦比香港多，而且外國的病人組織享有較多財力資助，並有很多興趣小組籌辦各種活動，例如籃球隊、游泳隊、羽毛球隊等，為年幼患者而設的課業輔導及遠足等社區活動、擇偶婚配的支援等也較多。

問起「小而同」，Tim稱讚道：「小而同成立的日子雖淺，但各方面的工作都做得不錯。」他將本港與外地的病人組織相比，認為「小而同」有很大機會在現有基礎上擴展。

對家長和教育工作者而言，Tim的事例讓我們看到，全人教育及關懷，對骨骼發育異常患者有重要意義。Tim回憶道：「家人的支持和愛最為重要。」他感謝家人、老師始終給予他信心，使他能在主流中學及大學完成學業。

A "little" pathology researcher who walked through pain and suffering

Jacqueline is a competent and intelligent young scientist at the Department of Pathology, The Chinese University of Hong Kong. She is currently a Post-doctoral research fellow focusing on genetics and cancer research, and has recently joined LPHK. She has been actively contributing to LPHK as a sharing speaker to motivate other "Little People", their parents and even university students.

Despite her success, Jacqueline has gone through a series of difficulties due to her condition. At birth, she was diagnosed with a rare type of growth hormone resistance which limited her skeletal growth and development. This condition is caused by a rare genetic mutation which occurs only 1 in 1,000,000 births, where the only treatment is surgical correction for structural problems and rehabilitation after surgery.

During her childhood, Jacqueline was offered an opportunity to have surgical correction by orthopedic surgeons. The purpose of the surgery was to lengthen and reconstruct her curved and deformed lower limb bones. At the time, her parents had opposing views on the surgery, with her mother believing that the surgery would offer a better quality of life for her daughter, while her father disagreed with having the surgery. Finally, her mother insisted on having the surgery, and so the surgeon proceeded after obtaining consent.

The surgery was successful and uneventful, but the rehabilitative period was far from an easy and comfortable one. After the surgery, Jacqueline had post-operative pain, wound infections and sleep deprivation. She missed many school classes and fell behind with her school work, and immobilization from surgery limited her socialization with classmates and friends. Her mother was blamed by her father and relatives at the time for making the decision to undergo surgery.

Jacqueline, however, appreciated that her mother made such a difficult choice for her when she was young. Despite the short-term problems, Jacqueline had suffered during the post-operative period, she benefited from a permanent increase in height and skeletal stability. Her mother was also very supportive in assisting Jacqueline with her rehabilitation program, which helped her overcome the difficulties she faced. For instance, her mother provided home-schooling to help her catch up with her school work.

Lower limb lengthening has enabled Jacqueline to carry out her daily living tasks and duties. For instance, she is now able to open doors and visualize objects on a table. She can also carry out her research experiments without help from others, and most importantly to her, see the delicious food on a dining table!

Retrospectively, Jacqueline is very grateful for her mother's insistence on surgery. Therefore, during every sharing session, Jacqueline always encourages parents to allow their children with skeletal dysplasia to undergo corrective surgeries if possible. Lastly, she also places great emphasis on family and social support, in addition to surgical correction. She believes that social support and acceptance are essential for children with skeletal dysplasia to be part of society. Without the support from her family and friends, she believes she could never have attained such a great height in her life, both physically and socially!

小個子，大作為

走出苦難的「小」病理學研究員

Jacqueline是一位聰明能幹的年輕科學家，目前在香港中文大學病理系擔任博士後研究員，專攻遺傳學和癌症研究，不久前更加入了「小而同」。她時常為「小而同」的成員及其家人以至大學生講述自己的經歷。

她的成功得來不易。Jacqueline經歷過一段艱難的日子。她出生時被診斷出患有一種罕見的疾病，稱為「生長激素抗性」，限制了她的骨骼生長和身體發育。這種疾病起因於一種罕見的基因突變，每一百萬新生嬰兒中只有一例。唯一的治療方法，就是作骨骼矯形手術和康復治療。

在Jacqueline小時候，骨科醫生說能為她作矯形手術，以拉長下肢骨骼，並矯正其彎曲變形。當時，父母對手術的意見不一，母親認為手術可能改善女兒的生活，但父親不同意。最後，在母親堅持下，骨科醫生為Jacqueline作了矯形手術。

手術十分成功，但康復時期不容易度過。手術後，Jacqueline須忍受疼痛、創口感染、睡眠不足等，錯過了許多課堂，學業成績落後了。手術後的調養，亦令她行動不便，減少與人交往。因此，她母親受到丈夫和親戚的指責。

但是Jacqueline知道母親為年幼的自己做了艱難的選擇，對她十分諒解。手術帶來短期的困難，但是手術永久地為她增加了身高，令骨骼穩定。母親也支持Jacqueline的康復治療，幫助她克服了許多困難，例如在家中給她補課，幫助她趕上學校的課業進度。

接受了下肢延長手術後，Jacqueline在日常生活中較容易做到了各種瑣事，例如能夠開門，看得見桌子上的東西。她還可以獨自進行研究實驗，不需要他人幫助。最重要的是，餐桌上有何美味食物，她能看到了！

回顧過去，Jacqueline很感謝媽媽堅持讓她做手術。因此，每次講述這番經歷，總是鼓勵家長儘可能讓骨骼發育不良的孩子接受矯正手術。最後，Jacqueline亦非常感謝家庭和社會的支持。她認為，身邊的人的支持和接納，對骨骼發育不良的兒童十分重要，是促使他們融入社會的第一步。她認為，若無家人和朋友的支持，自己今天的身型及在社會上的位置不會有如此「高度」！

From bedside to operating theatre - nothing could hold him back

As a pediatric orthopedic surgeon at Johns Hopkins Hospital, Dr Michael Ain has come a long way to arrive at where he is today.

After graduating with a Math major from Brown University, Dr Ain decided he wanted to be a medical doctor. Despite his excellent academic results and impressive list of extra-curricular activities at Brown, rejection letters from the 20 to 30 medical schools he had applied to arrived one after another. "I was scared. I was angry. I was hurt. It goes against everything I was taught. My parents told me I could do anything I wanted as long as I pushed myself," he recalled. Although none of the letters mentioned his height, he had no doubt that some or all of the admissions officers believed a dwarf could not become a physician.

Determined to pursue his dream, Dr Ain returned to Brown and took two advanced science courses, continued his research and published his work. He applied for 20 medical schools again the next year and eventually got accepted into Albany Medical College. It was at Albany that he decided to become a pediatric neurosurgeon, and subsequently an orthopedic surgeon.

Dr Ain had not considered specialising in bone disorders until he heard a desperate couple with an achondroplastic daughter said they had regained hope after meeting Dr Ain. "I thought, if I could help people, or be able to sympathise and understand certain issues, then maybe that's why God wanted me to become an orthopedic surgeon," said Dr Ain. He then applied for a fellowship in orthopedic surgery at Johns Hopkins Hospital. This time, the director of orthopedic surgery had no hesitation about hiring Dr Ain because of his proven capability at Albany.

Today, Dr Ain treats children with various types of skeletal dysplasia, including those who have the same condition (achondroplasia) as himself. For the children who come to his clinic, he is not only a doctor, but a role model who shows them they can do anything they want as long as they are determined.

小個子，大作為

從病床到手術室 — 困難無法阻擋他

Michael Ain 醫生是美國約翰．霍普金斯醫院的兒童骨科醫生，走過了漫漫長路，才來到今天。

Ain醫生在美國布朗大學數學系畢業後，立志從醫。雖然他在布朗大學的成績優秀，並積極參與課外活動，但他報讀的二三十所醫學院陸續寄來覆函，沒有一所願意錄取他。Ain醫生回憶道：「我很害怕，很憤怒，很傷心。這跟我歷來學到的完全相反：父母告訴我，只要我努力，想做甚麼都可以做到。」雖然沒有一封覆函提及他的身高，但他肯定有些招生人員認為，甚至全部收生人員都認為，侏儒症患者不能成為醫生。

Ain醫生立志追尋夢想，就回到布朗大學，修讀兩個高級的科學課程，繼續研究工作，發表成果。第二年，他報讀20所醫學院，最終獲奧爾巴尼醫學院錄取。在奧爾巴尼醫學院求學期間，他決定做一個兒童神經外科醫生，後來決定做矯形外科醫生。

Ain醫生沒想過要專研骨骼異常疾病，直至他聽到一對夫婦說起遇見他之後重拾對侏儒症女兒的希望。Ain醫生說：「我就想，如果說我能夠幫助人，能夠同情、理解某些問題，那也許因為是上帝有意要我做一個矯形外科醫生。」後來他向約翰．霍普金斯醫院申請一個矯形外科研究員職位。這次，矯形外科的主管毫不猶豫聘用了Ain醫生，因為他已在奧爾巴尼醫學院證明了自己的能力。

今天，Ain醫生醫治患有各類骨骼發育異常病症的兒童，包括跟他同樣患軟骨發育不全症的孩子。對於來到他診所的兒童，他不只是一位醫生，也是一個榜樣，讓孩子們看到：只要有決心，無論想做甚麼，都可以做到。

A Chair Professor who advocates human rights for the disabled

Professor Sherry Chen is a successful Chair Professor at the Graduate Institute of Network Learning Technology at Taiwan National Central University.

Born with achondroplasia, Sherry recalls facing numerous biases and discrimination in her life. When she was in primary and secondary school, her classmates often teased her about her condition and bullied her. However, instead of giving up on herself, she devoted her energy and effort to her studies. Her academic excellence ultimately led her onto a path of academia and research. Sherry obtained her Bachelor's degree in Library and Information Science from The Taiwan Fu Jen University and her Master's degree from The University of Maryland in the United States. She subsequently returned to Taiwan and started her career at The Legislative Yuan of Republic of China at the age of 26.

Despite her smooth academic and career path, she was not satisfied with her life. She lamented that her short stature limited her from having a romantic relationship, not to mention starting a family. Facing this adversity, she again converted her worries into motivation, and applied for a PhD at the University of Sheffield. She focused on her research interests in collaborative and multimedia learning, and other digital influences in our daily lives. She has now published more than 150 research papers in this field, and was granted an Outstanding Scholar Award by the Foundation for Advancement of Outstanding Scholarship in Taiwan in 2010. Retrospectively, she attributed her success to her singular focus and being free from hustle and bustle of having a family.

In addition to her academic contribution, Sherry is also an advocate of human rights for disabled people. Understanding the plight and discrimination a disabled person may experience in modern society, Sherry is actively involved in giving motivational talks to inspire others to strive for excellence in life. In recognition of her contribution to uphold the rights of disabled people, she was awarded The Caring for Life Award in 2012 by the Dharma Drum Mountain Humanitarian and Social Improvement Foundation.

小個子，大作為

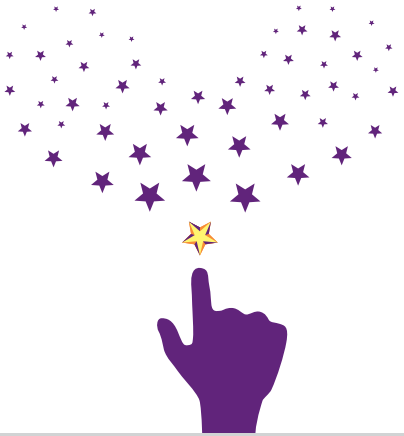
捍衛殘疾人士權利的講座教授

陳攸華 教授是台灣國立中央大學網絡學習科技研究所的講座教授。

陳攸華 教授生來就有軟骨發育不全，回憶中難忘年幼時在生活中曾遭遇許多偏見和歧視。上小學和中學時，同學經常因她的病情而嘲笑、欺負她，但她不自暴自棄，只把全副精力傾注在學習上。她的學業成績優異，從此走上學術研究之路。她從台灣輔仁大學圖書資訊學系獲得了學士學位，又從美國馬里蘭大學取得了碩士學位。後來26歲時從美國回台灣，任職於中華民國立法院，開始了她的職業生涯。

儘管在學術和職業生涯中一帆風順，她對生活未能滿足。她慨嘆自己因身材矮小所限，無機會戀愛，更談不上成家和生兒育女。面對逆境，她再次把憂慮化為動力，申請前往英國謝菲爾德大學攻讀博士學位。她的研究專注於協作式學習、多媒體學習，以及數碼科技在當今生活中的影響。她現已發表了150多篇該領域的研究論文，並在2010年獲得台灣傑出人才發展基金會頒授的傑出人才獎。回顧過去，她說自己的成功可歸因於專注做一件事，沒有家庭帶來的煩惱。

除了對學術有貢獻，**陳攸華** 教授亦是殘疾人士的人權倡導者。她本身患有軟骨發育不全，更能理解當代社會中殘疾人士面對的困境和歧視。她積極參與勵志講座，鼓勵殘疾人士追求卓越的人生。為表彰她保障殘疾人士人權的貢獻，法鼓山人文社會基金會在2012年向她頒授「關懷生命獎」！



Perseverance built on faith and action

> *"They will say you are on the wrong road, if it is your own."*
>
> *Antonio Porchia*

Josie obtained her Master of Information Technology Management at The Chinese University of Hong Kong. Subsequently, she and her friends founded a company which develops software for use in the fashion industry. The company joined the 2-year Cyberport Creative Micro Fund Scheme last year, and expects to have 280,000 clients by 2017.

Josie identified her strengths through other people. Facing the physical limitations of dwarfism, Josie admitted she could either accept her restrictions or insist in a breakthrough - she chose the latter. "I believe as long as I can enjoy the same rights and happiness as normal people, I cannot accept an attitude of 'this is all you can do'". However, she believes that perseverance also requires faith and action, and that being foolhardy should not be seen in a negative light. "When you want to prove you are right to others, you have to take action. Actions speak louder than words, and you need to try different methods to think outside the box", which she considers is better than not taking any action at all. While some mistakes may be made in the process of 'try and err', lessons will be learned and mistakes corrected with each attempt.

Josie's greatest challenges are those associated with her physical restrictions. The first time she went abroad alone was when she was 19 and her biggest worry was how she would carry her luggage, which was almost the same height as her shoulder. After that trip, she realised that she found something difficult, only because she mentally thought it to be, and that it helped to see

things from a different perspective. During a recent trip to Japan, Josie described the willingness of strangers to help if she asked for it. "It is not because you have short stature, but because they are kind. What you need to do in return is to appreciate and be thankful for their time and kindness".

Josie shared that "I accept problems derived from my limitations, and try to learn how to do better. Those around you will offer help and share their experiences when they recognize your persistence. This allows you to do better". At a younger age, Josie felt humiliated and embarrassed when teased by other children. Today, she understands those children laughed because they saw someone who was different, not because they wanted to tease her. It is true that few parents in Hong Kong know how to deal with such situations. In such instances, Josie encourages Little People to start a conversation and communicate, as this is an opportunity to educate them and let them know that being different is a human characteristic.

Message to Little People

There are two messages Josie would like to share with Little People: firstly to accept their own limitations, and secondly to live a happy life. These two ideas are closely linked - you need to accept your limitations before you can enjoy true happiness.

小個子，大作為

堅定信念，堅持行動

「如果那是你自己的路，人家會說你的路走錯了。」

安東尼奧 • 波契亞

譚慧怡(Josie)是中文大學資訊與科技管理學碩士。她與朋友創辦了一家公司，為時裝業界開發軟件。公司去年加入為期兩年的數碼港培育計劃，期望到2017年能有28萬名用戶。

Josie說通常是身邊的人指出她的優點。面對侏儒症在體格上的限制，Josie坦言只有兩個選擇：甘心接受限制，或堅持打破限制，而她選擇了後者。「我認為只要能和其他人一樣享有權利、一樣快樂，我就不甘心接受別人說的『你就只能做到這樣』。」她的堅持，源於自己的信念和行動。她不同意「有勇無謀」是壞事，總好過無勇又無謀。她說：「你要向別人證明自己是正確的，就必須有行動，不能只說不做，必須想方設法打破框框。」在『試錯』過程中，必定會犯錯，但可以從錯誤中學習，改正錯誤，取得教訓。

Josie最大的困難是受體格所限，有些事情做不到。她第一次獨自出國是19歲那年，當時她最擔心的是：行李箱幾乎高到肩膀，如何搬得動？但那次旅程之後，她認識到有些事覺得困難，只是自己的看法而已；能夠從不同角度看事情，問題便可解決。去年Josie到日本參展，她說她會主動開口請人幫助，「人家都很樂意幫忙，不是因為看你身材矮小，而是因為他們善心。你只需要對他們付出的時間和友善表示欣賞和感謝。」

Josie繼續分享說：「我接受局限帶來的問題，努力學習如何改進。身邊的人看見你堅持不懈，會給你幫助，並交流他們自己的經歷，使你能做得更好。」她有時會被小孩子取笑身材矮小，年幼時她會感到難堪和羞辱，但她現在學會了接受，明白小孩子是因為看見別人不同於自己而笑，而不是惡意取笑。的確香港仍有很多父母不太懂得應對這種情況。Josie鼓勵侏儒症患者遇到這境況時更應該主動與對方交談、溝通，藉此機會讓小孩子明白身高、體重的不同，是他人的特徵。

寄語「小個子」

Josie有兩個忠告想送給各位「小個子」：一是要接受自己的局限，二是要生活得快樂。兩個意念，彼此相連：先要接受局限，才能生活得快樂。

Useful resources for parents and teachers

General information

Common types of skeletal dysplasia
http://www.lphk.org/SkeletalDysplasia.html

First steps for parents:
"It's A Whole New View; A Guide for Raising a Child With Dwarfism"
http://www.lpaonline.org/for-parents-and-teachers

For children with achondroplasia:
"Health Supervision for Children With Achondroplasia"
http://www.lpaonline.org/for-parents-and-teachers

Adaptive products
http://www.lpaonline.org/adaptive-products-

Home and school modifications

Ideas for home adaptations
http://www.lpaonline.org/a-guide-to-home-modifications

Ideas for school modifications
http://www.lpaonline.org/for-parents-and-teachers

Education

Parenting and children books
http://www.lpaonline.org/parenting-and-childrens-books

給家長和教師的相關資訊

一般資料

常見的骨骼發育異常
http://www.lphk.org/SkeletalDysplasia.html

給家長的第一本指南：
《這是新角度：養育侏儒症孩子的指導》
http://www.lpaonline.org/for-parents-and-teachers

患軟骨發育不全症的兒童：
對患軟骨發育不全症兒童的健康監察
http://www.lpaonline.org/for-parents-and-teachers

協助適應生活用品
http://www.lpaonline.org/adaptive-products-

家居及學校的改建

家居改建的建議
http://www.lpaonline.org/a-guide-to-home-modifications

學校改建的建議
http://www.lpaonline.org/for-parents-and-teachers

教育

育兒及兒童書籍
http://www.lpaonline.org/parenting-and-childrens-books

About Little People of Hong Kong

Little People of Hong Kong (LPHK) Foundation was established in 2013 and strives to help rare bone disease patients to live an independent and fulfilling life by overcoming challenges associated with their physical disabilities.

We achieve our mission through:

- Establishing a support group which enhances the physical, psychological and social well-being of patient families;
- Providing useful and timely medical information to patient families;
- Promoting social acceptance of patients by educating the public about rare bone diseases.

LPHK is primarily an all-volunteer organization operated by an Executive Committee supported by an Advisory Board which consists of physicians, professionals and patient's families

Website: http://www.lphk.org
Email: info@lphk.org
Facebook: www.facebook.com/littlepeoplehk

關於小而同罕有骨骼疾病基金會

「小而同罕有骨骼疾病基金會」於2013年創立，旨在協助患罕有骨骼疾病的人士克服身體殘障帶來的困難，從而幫他們爭取獨立而有意義的生活。

我們以如下方式實現使命：

- 建立病人支援小組，以助增進患者及其家人的身心健康，幫助他們融入社會；
- 為患者家庭適時提供有用的醫藥護理資訊；
- 為大眾傳播骨骼發育異常的知識，以期促使社會接納此類疾病的患者。

「小而同罕有骨骼疾病基金會」的運作由執行委員會管理，而顧問委員會則由醫護人員、專業人士及病人家屬組成，所有人員均屬義工。

網址: http://www.lphk.org
電郵: info@lphk.org
Facebook：www.facebook.com/littlepeoplehk

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Skeletal dysplasia is a general term referring to the abnormality of bone growth. In many cases, skeletal dysplasia will result in extreme short stature which is commonly known as dwarfism. Due to their extreme short stature, people with dwarfism are referred to as "Little People" in many parts of the world. In this book, we focus on the medical, emotional and social challenges Little People face from birth to adulthood, using real-life examples to show how they, with the support of family and society, overcome those challenges to live a fruitful life.

「骨骼發育異常」泛指骨骼生長不正常所致的一類疾病，往往導致患者的身材特別矮小，也即患上俗稱的「侏儒症」。由於身軀特別矮小，在很多國家中，這類患者經常被稱為「小個子」。在本書裡，我們集中討論「小個子」從出生至成年所面對的種種挑戰，包括治療、情緒、社交等各方面的難題，並以真實的事例展示「小個子」怎樣獲得家庭及社會的支持而克服困難，活出豐盛的人生。

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