



Sickle cell disease: a lethal advantage

Transmitting genetic insights

SUSAN RAE

As in the Philadelphia, education of parents about sickle cell disease is a central part of the programme.

JEMIMA DENNIS-ANTWI

I do genetic counselling in terms of trying to explain to the parents how come we are telling them the child has sickle cell disease. So we have some little boxes we call inheritance boxes. In these boxes we have four tennis balls, we show the various probabilities. For example we decide that you are AS and your husband is also an AS. You want to have less than six children. Let's see the outcome of each pregnancy so they pick and up it comes on the tennis what we've written, the various probabilities. And they seem to understand, oh that's why maybe my first which hadn't the disease but this third one had it or maybe why my first baby had it.

Traditionally mothers are most always involved in taking care of their child. We tell them that this is why you have come to the clinic. This is the programme we are carrying out and we have found out that your child has possible sickle cell disease. And it is not through you but it is through yourself and your husband that the child caught this disease. And most often they go home to tell their husbands and some of them would say, I mean it's not true, I'm not sick, I've never had any disease, I mean I've never shown any symptom of the disease, so it is not true. They refuse to accept the fact that they have this child with sickle cell disease. Sometimes they refrain their wives from coming to the clinic.

SUSAN RAE

Getting across the message about the true nature of sickle cell disease to fathers and the public in general involves a variety of media, including talk shows on the radio, poetry and song.

KO NIMO

Some times when you have a child with this disease, parents don't usually combine efforts in helping, and some instance where after a fight the man would say, oh you inherited that from your mum. They forget that it's the two of them. So I wrote this little song and put in ideas like 'daddy and mum, when I was born. I was quite a heavy child, I brought happiness into the house, and now why have you even decided not to take care of me because I get ill often.

SUSAN RAE

Hannah and Mary are community nurses attached to the new born screening project. Their task is to track down the parents of babies that have been diagnosed as SS and get them to enrol in the clinic and education programme. They also have to follow up patients that have been enrolled at the clinic and then failed to show up. In a city where few people have a telephone this means searching out families in their communities on foot. Despite the lack of accurate addresses or reliable landmarks, the nurses still manage to locate most of the families they're seeking.

MARY LAMPTEY

The person said, they put a landmark - it's moon light, moon light and sevens, it's what the person said. Unfortunately for me the house we located is no where near the important landmark.

SUSAN RAE

In Philadelphia, social worker Chuck Adams also has to deal with the wider social problems faced by sickle cell patients and their families.

CHUCK ADAMS

The disease itself makes it very difficult often times to fit a solution into a problem. With a cold day like today, this family that's living in an abandoned building would have to be taken to a shelter, but they would have to be taken to one that has medical support. If we have a family that has general heating problems, no food, situations like that, all of this will have an affect on a family with children with sickle cell disease. They just happen to have a chronic genetic disorder, but being poor was probably the first disorder that they had to deal with.

SUSAN RAE

The prognosis for children that are diagnosed with sickle cell disease at birth and enrolled in a comprehensive treatment programme like that at the Children's Hospital of Philadelphia is constantly improving. Many sickle cell specialists now see the main challenge as increasing awareness of the disease in the community at large.

KIM SMITH-WHITLEY

Over the course of the years sickle cell disease has almost been an orphan disease in the sense that it's affected primarily African Americans, which historically has had a very turbulent time in the United States. A lot of individuals with sickle cell disease were dying early. I think as the numbers grow, as we identify more new-borns with sickle cell disease, one in every three hundred and seventy five African American new-borns has sickle cell disease.

Hopefully those children will become teenagers, become young adults, and if we as physicians start educating not only the child, but educating the child's community, then multiple communities will be educated as well as multiple individuals. And I think that's the only way communities are going to realise what the availability is for pre-natal diagnosis, what the availability is for cures like gene therapy hopefully in the future or bone marrow available for individuals with sickle cell disease. We really have to start by educating the entire community, and you can't do that until physicians empower their patients transplantation, or realise what palliative therapies like blood transfusions or hydroxyurea are.