RARE DISEASES AND HOMOEOPATHY

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Rare Diseases and Related Terms

Rare diseases terms are either terms for which information requests have been made to the Office of Rare Diseases Research, the Genetic and Rare Diseases Information Center, or the National Human Genome Research Institute; or diseases that have been suggested as being rare. The purpose of the Rare Diseases and Related Terms list is to distribute information; although the list is updated regularly, it should not be used as a reference or guarantee that a condition is rare. The prevalence of a rare disease is usually an estimate and may change over time. A rare (or orphan) disease is generally considered to have a prevalence of fewer than 200,000 affected individuals in the given country.

Let me begin with an interesting case of Wilson's disease. I will first introduce a small synopsis of the disease.

WILSON'S DISEASE

Wilson's disease is a rare inherited disorder that causes copper to accumulate in the liver, brain, and other vital organs. In individuals with Wilson's disease, copper is not eliminated properly and instead accumulates, possibly to a life-threatening level. Left untreated, Wilson's disease is fatal. When diagnosed early, Wilson's disease is easily treated, and many people with the disorder live normal lives. Copper can accumulate in and damage the liver.

Because copper first accumulates in the liver, most individuals with Wilson's disease initially have signs of liver damage, including abdominal pain and yellowing of the skin and whites of the eyes (jaundice).

Causes

In Wilson's disease, a genetic mutation affects ATP7B, a protein that helps transport copper into the bile. ATP7B is also involved in incorporating copper into ceruloplasmin, a protein that carries the mineral through the bloodstream. The defects in the ATP7B gene mean that copper is not eliminated properly, and instead builds up in the liver, where it can cause serious and sometimes irreversible damage.

Risk Factors

If both parents are carriers of one abnormal Wilson's gene, they have a 25% chance of having a child with two normal genes, a 50% chance of having a child who also is a carrier, and a 25% chance of having a child with two recessive genes who will develop the disease. These chances are the same in each pregnancy.

Signs and Symptoms

In people with Wilson's disease, copper begins accumulating in the liver immediately after birth, but signs and symptoms rarely occur before the age of five or six and sometimes not until ages 40-50.

The most characteristic symptom of Wilson's disease is the Kayser-Fleisher ring - a rusty brown ring around the cornea of the eye that can best be viewed using an ophthalmologist's (eye doctor) slit lamp. The primary consequence for most of those with Wilson's disease is liver disease, appearing in late childhood or early adolescence as acute hepatitis, liver failure, or progressive chronic liver disease in the form of chronic active hepatitis or cirrhosis of the liver.

The main symptom of liver disease is jaundice. Jaundice is the yellowish staining of the skin and sclerae (the whites of the eyes) that is caused by high levels of the chemical bilirubin in blood. Bilirubin is a brownish yellow substance found in bile. It is produced when the liver breaks down old red blood cells. Bilirubin is then removed from the body through the stool (feces) and gives stool its normal brown color. The color of the

skin and sclerae vary depending on the level of bilirubin. When the bilirubin level is mildly elevated, they are yellowish. When the bilirubin level is high, they tend to be brown. Icterus is the term for yellowing of the sclerae.

Other signs and symptoms of liver toxicity include: abdominal pain and swelling; chronic itchy skin; dark urine color; pale stool color; joint pain; bloody or tar-colored stool; chronic fatigue; nausea; loss of appetite; fatty liver, fibrosis, cirrhosis, and liver failure.

In others, the first symptoms occur later in adulthood and most commonly include slurred speech (dysarthria), difficulty swallowing (dysphagia), and drooling. Other symptoms may include tremor of the head, arms, or legs; impaired muscle tone, and sustained muscle contractions that produce abnormal postures, twisting, and repetitive movements (dystonia); and slowness of movements (bradykinesia). Individuals may also experience clumsiness (ataxia) and loss of fine motor skills.

A third of those with Wilson's disease will also experience psychiatric symptoms such as an abrupt personality change, bizarre and inappropriate behavior, depression accompanied by suicidal thoughts, neurosis, or psychosis.

Complications

Wilson's disease can increase the risk of bone fractures (osteoporosis) of serious infections and may greatly impair kidney function. But one of the most serious complications is liver damage, which may be so severe that only a liver transplant can prolong life. If not treated, Wilson's disease is fatal.

Diagnosis

Blood tests:
A combination of blood tests will be used to determine if Wilson's disease is present.

Genetic tests: Because more than 200 mutations of ATP7B exist, researchers have not been able to develop a simple

genetic test that can help screen or diagnose Wilson's disease in the general population. However, a procedure called haplotype analysis can identify people within a single family who may have inherited the disorder.

Copper levels: Levels of copper in the blood will be determined with a blood test.

Albumin: Serum albumin levels measure the main protein made by the liver and reveal how well the liver is making this protein.

Bilirubin: Bilirubin is a waste product made from old blood cells; it is a yellow compound that causes jaundice and dark urine when present in increased amounts. Tests for bilirubin levels help determine if the liver is functioning appropriately.

Liver enzymes: Another blood test may be performed to check for elevated levels of liver enzymes, such as alanine aminotransferase (ALT) and aspartate aminotransferase (AST). These enzymes leak into the bloodstream when liver cells are injured. Also, alkaline phosphatase (ALP) levels may be checked. ALP is an enzyme related to the bile ducts. ALP levels are often increased when they are blocked.

Total protein: Total protein tests measure albumin and all other proteins in blood, including antibodies made to help fight off infections.

Liver biopsy: A liver biopsy may be performed to determine the extent of liver damage and to determine the best treatment option for the patient. During the procedure, a needle is inserted into the liver and a small tissue sample is removed. The tissue is then analyzed under a microscope in a laboratory.

CASE:

In late 80's I was invited in the city of Indore, Madhya Pradesh to see a patient 19 old, suffering from early onset Wilson's disease with cirrhosis of liver, jaundice, liver failure and early kidney failure. He also was suffering from early congestion in the lungs with pneumonia. The case was being treated

adequately with allopathic medicine but the child's unconsciousness was gradually progressing and the child was going into a deeper and deeper hepatic coma.

I started noting down all my observations that I could make. I saw that the patient was emaciated, icteric and was mildly delirious along with this there was moaning and muttering. There was a low grade fever of 99° F - 99.6 F°. As the evening and night approach the delirium became more aggravated. When not in delirium the person is quite indifferent, lying quiet without asking any food or water. The speech was little low volume and occasionally incoherent. There was mild degree of photophobia. The expression of the face was sickly, there was cold perspiration on the face, the tongue was discoloured brown especially in the centre, offensive odour was coming from the mouth, trembling of the tongue when protruding, complete absence of appetite, and there was extreme thirst of small quantity. The person will periodically vomit small quantities of water drank and the food eaten.

When I touched the abdomen it was cold to touch. Parents complained that he was passing brown coloured highly offensive stool but slowly the stool is becoming more light yellow. The urine was yellowish reddish and dark. There was a cough on attempting to talk, the back was stiff, the skin was dark yellow, the blood report shows anaemia, bilirubin of 12 mg, SGPT of 550 IU, the pulse was small, tremulous and weak. The extremities showed trembling of the hands. There was excessive weakness the person was hardly able to walk.

I had a long discussion with the parents regarding the prognosis the parents agreed to that and requested me if only I can give relief in jaundice that would be great, as due to jaundice he was constantly scratching the skin that was dry. He was not eating at all, he was vomiting and he was delirious. The parents wanted me to treat those symptoms symptomatically with the correct indicated remedy. I asked the past history and the parents said that he was a very intelligent student but he always wanted to join the science faculty after his SSC, unfortunately he had malaria during his SSC examinations and as a result his performance in exams was not so good and he could not get admission in the science faculty

and as a result he was given an admission in a commerce faculty. And that was the big grief in his mind because the expectations were too much from his friends and family and he could not fulfill the expectations and since then he became depressed and dejected.

He was studying it without any ambition, he always remained sad and depressed since then, and slowly he observed that he is developing tremors in his extremities while lifting the things and writing and during any exertion. Then he complained of distension of abdomen, constipation, dyspepsia and slowly he started developing nausea and vomiting with no appetite.

When he was investigated with all these procedures the diagnosis of Wilson's disease came. Later on when the doctors inquired in the hospital they informed that the grandfather too had suffered from jaundice they were not sure whether it was Wilson's disease or not.

After the SSC examination the person in his depression was quite lazy and he also had weak memory especially making mistakes in spelling, words and names of the persons, etc…

Keeping all this information in account I prescribed him Crotalus horridus 200C, every 3 hourly diluted in a glass of water one teaspoonful and I asked them to continue this for 4 days.

When I came back to Bombay I was informed that the vomiting which used to happen at least 20-25 times a day had lessen down to 15 times a day, nausea was better, the appetite was little improved. So I asked them to continue for 7 more days. Gradually within 15 days the vomiting completely disappeared and the nausea was mild, appetite still not returning to normal. I asked them to perform a blood test which shows bilirubin 8 mg, a definite drop of 4 mg in 7 days. The heamoglobin which was 10.7 gm% jumped up to 11.3 gm% which was also a good sign. I continued the same medicine for 3 more weeks, slowly over a period of 2 months the bilirubin came down to 5.7 mg, the SGPT came down to 220 IU and there was no vomiting.

At the completion of 3 month of homoeopathic treatment the patient was completely asymptomatic, only the liver scans the sonography showed cirrhosis of liver and there was a trembling which never came under control with Crotalus horridus.

After the stage of Crotalus horridus was over I tried to focus more on the trembling, the cirrhotic pattern and his extreme degree of thirst. Now I prescribed him Phosphorus 12C every 4 hourly for 10 days and I asked them to repeat the liver function test which clearly showed that the bilirubin dropped down to about 4.6 mg. the SGPT came down to 150 IU, the appetite was normal and there was much more level of comfort soon after starting Phosphorus 12C.

I asked the patient to continue Phosphorus 12C three times a day for a period of 2 months and with this I could keep the complication of cirrhosis much more under control for the next 4 months. So in totally 6 months of homoeopathy, first 2 months with Crotalus horridus and then followed by Phosphorus, I was able to palliate this case. Later on I heard that the patient developed an intercurrent gram negative infection, was hospitalized where he succumb to death. However for 6 months with such a rare disease I could control the symptoms with Crotalus horridus and Phosphorus.

PRADER-WILLI SYNDROME

Prader-Willi syndrome (PWS) is a genetic disorder that occurs in approximately one out of every 15,000 births. PWS affects males and females with equal frequency and affects all races and ethnicities. PWS is recognized as the most common genetic cause of life-threatening childhood obesity.

What are the symptoms of Prader-Willi syndrome?

The symptoms of Prader-Willi syndrome are thought to be caused by dysfunction of a portion of the brain called the hypothalamus. The hypothalamus is a small endocrine organ at the base of the brain that plays a crucial role in many bodily functions, including hunger and satiety, temperature and pain

regulation, fluid balance, puberty, emotions, and fertility. Although hypothalamic dysfunction is believed to lead to the symptoms of PWS, it is unclear how the genetic abnormality causes hypothalamic dysfunction.

There are two generally recognized stages of the symptoms associated with PWS:

Stage 1

In the first stage, infants with PWS are hypotonic or "floppy", with very low muscle tone. Weak cry and a poor suck reflex are typical. Babies with PWS usually are unable to breastfeed and frequently require tube feeding. These infants may suffer from "failure to thrive" if feeding difficulties are not carefully monitored and treated. As these children grow older, strength and muscle tone generally improve. Motor milestones are achieved, but are usually delayed.

Stage 2

An unregulated appetite characterizes the second stage of PWS. This stage most commonly begins between ages 2 and 6 years old. Individuals with PWS lack normal hunger and satiety cues. They usually are not able to control their food intake and will overeat if not closely monitored. Food seeking behaviors are very common. In addition, the metabolic rate of persons with PWS is lower than normal. Left untreated, the combination of these problems will lead to morbid obesity and its many complications.

In addition to obesity, a variety of other symptoms can be associated with Prader-Willi syndrome. Individuals usually exhibit cognitive challenges with measured IQs ranging from low normal to moderate intellectual disability. Those with normal IQs usually exhibit learning disabilities. Other issues may include speech apraxia/dyspraxia, short stature, small hands and feet, scoliosis, sleep disturbances with excessive daytime sleepiness, undescended testicles in males, high pain threshold, and infertility. Behavioral difficulties may include obsessive-compulsive symptoms, skin picking, and difficulty controlling emotions. Adults with PWS are at increased risk for

mental illness. PWS is a spectrum disorder and symptoms vary in severity and occurrence among individuals.

CASE:

This is the case of a child who was complaining of Prader-Willi syndrome. Now this child basically was brought to me at the age of 9 years old by the mother who was a single mother she was single since the past many years but the child had an equally good contact with the father also, so for some days the child used to stay with father and for some days the child used to stay with the mother. Also mother being a business woman and very busy professional most of the time the child used to stay with the grandmother.

Now the chief complaints when the child was brought to me were basically obesity and behavioural problem. The child went to a special school but the child had a tremendous appetite, will eat, eat, and eat whole day. And along with that the child will also have lot behavioural problem - Compulsive obsessive behaviour, the child will do certain things in a certain way only and will not understand whatever the explanation is given to the child in relation to that.

Food wise she had strong cravings for chocolates, cold drinks, onions, salt, sweets and tea. She had sticky, greasy and offensive perspiration on the body. She was little timid in public places. Intellectually she was not very sharp. Her whole focus of attention was basically on eating to survive, I can say that.

She had dark hair, she cannot undertake the mental stress, and now living with single parent definitely she had lot of conflicts in her mind regarding her relationship with the parents. She sometimes complained of catarrhal headache, her nasal catarrh will be yellowish greenish; she also has a past history of otitis media. Her stool were little greasy and fatty. She was a hot patient; she never tolerated heat in any form. I interviewed the mother and father to know more something about the characteristic feature of the child.

Both the parents came to a common conclusion that the child has ravenous appetite of a fixed way of living the life, fixed way of dressing, a fixed way of selecting the cloth. She was extremely childish retarded with a strong craving for chocolate, sweets and tea.

With these symptoms I prescribed her Thuja 30C, I observed the child for the period of next 2 years where I was able to control the child's appetite. The persistent thought that is the compulsive behaviour, the obstinacy in the child, child could focus better in the classroom.

From 30C I went to 200 and later 1M potency. Knowing fully that Prader-Willi syndrome cannot be cured with homoeopathy, these are the area where the homoeopathic doctor should attempt especially the appetite, the obesity and the compulsive obsessive behaviour.