

**NIEMANN-PICK TYPE C, A RARE GENETIC DISORDER, AFFLICTS AN 8-YEAR-OLD**

# Aaditya's silent suffering



**FAMILY SUPPORT:** Aaditya with his mother Tasneem and father Ravi at their residence, in Bangalore

**Soumita Majumdar, BANGALORE**

Like any child his age, Aaditya loves to watch his favourite cartoon show on television. But the eight-year-old knows he is different as he cannot walk. Talking and eating are even more laboured efforts.

Aaditya has been suffering from a condition known as Niemann-Pick Type C (NPC) since his childhood. Also known as Alzheimer's in children, the disease is caused by genetic mutation. It leads to lipid and cholesterol accumulation in the cells which then start dying, affecting various organs like the brain, the liver and the spleen.

"Till three years of age, Aaditya was a normal child. He had even started going to school. One day, we noticed that his spleen was enlarged. But we thought it was normal. Later, he developed Vertical Gaze Palsy, where the person looks upwards. We knew something was wrong somewhere," says Tasneem Dasgupta, his mother. For Aaditya's parents, the struggle had only begun.

"While his symptoms became apparent over a period of time, we took him to the best of doctors all over the country. But they failed to diagnose Aaditya's problem," says Ravi Dasgupta, his father. "NPC is a rare disease. But doctors in India are still unaware of it. Aaditya's test results showed he was normal, but again, we knew, something was terribly wrong.

**SO WHAT IS NIEMANN-PICK TYPE C (NPC)?**

**NPC is a lysosomal storage disease** associated with mutations in NPC1 and NPC2 genes

**NPC strikes** an estimated 1:150,000 people, according to The Journal of B Chemistry

**Approximately 50 per cent** of cases present before 10 years of age, but manifestations may first be recognised as late as the sixth decade

**There is no known cure** for NPC, nor

is there any FDA-standard approved disease modifying treatment.

**Supportive care** substantially improves the quality of life

**Children with** antenatal or infantile onset usually succumb in the first few months or years of life, whereas adolescent and adult onset forms of NPC have a slower progression, and individuals may survive to the seventh decade

**Tasneem Dasgupta**

Aaditya's mother

**We noticed that his spleen was enlarged. We thought it was normal. Later, he developed Vertical Gaze Palsy, where the person looks upwards. We knew something was wrong somewhere."**

After coming across symptoms of NPC that were similar to Aaditya's problem, we took him to Gangaram Hospitals in Delhi, the only place where skin biopsy is done for screening NPC," says Dasgupta. Samples sent to the Netherlands for tests took nine long months before confirming Aaditya's case as one of NPC.

Till date, there have not been any treatments available for NPC. There is, however, an experimental drug available in Australia, costing around A\$7000 for a dose of 45 days that helps slow down the progression of the disease.

An NPC victim needs to be on this drug all his life. "It was never an option as the medicine was way beyond our capacity," says Tasneem. So far, Aaditya's parents have spent Rs8 lakhs on his treatment.

Recently, Tasneem and Dasgupta thought they saw a ray of hope. Aaditya was the first person to have undergone intra-cranial stem cell transplant in one of the city's super-specialty hospitals in December, 2008.

Done on an experimental basis, only time can prove its success.

In NPC, the brain cells become damaged. Mesenchymal cells have been injected into Aaditya's brain through the skull. A stem cell transplant generates new neurons that can restore the functions of the brain. "Soon after the treatment, he showed signs of improvement, though it is too early to say anything," says Tasneem. But a bout of pneumonia led to further deterioration of the child's condition.

Worldwide, only 500 children are known to be affected by NPC. Statistics remain unrecorded in India on the number of NPC cases. "There may be more cases. But since doctors are in the dark about NPC and its symptoms, diagnosis remains a problem," says Dasgupta.

In order to educate people about NPC, Aaditya's parents have set up a website called www.hopeforaaditya.com. "We want to generate awareness about NPC among more people. What we went through while being ignorant about the disease was frustrating. Through this website, we can network with parents of other affected children all over the world, share medication details and day-to-day experiences," says Dasgupta.

**askthedoctor**

## LUMP on your neck? Could be thyroid cancer

**T**h thyroid cancer occurs when the cells of the thyroid gland grow uncontrollably to form tumours that can invade the tissues of the neck, spread to the surrounding lymph nodes, or to the bloodstream and then to other parts of the body. Different cancers develop from different kinds of cells in the thyroid gland, which in turn determine the type of treatment needed.

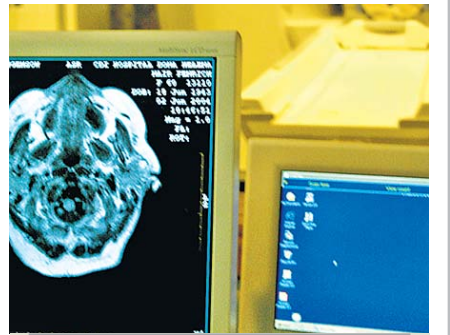
**WHAT ARE THE SYMPTOMS?**

As the thyroid gland is close to the skin, tumours often appear as bumps in the neck, and is by far, the most common presentation. They are called thyroid nodules. As the thyroid gland is a fairly superficial organ in the neck, a thyroid nodule can be noticed during its early stage, and medical attention be sought immediately. A change of voice is also an indicator of thyroid cancers with symptoms such as shortness of breath, hunger, difficulty or pain while swallowing, or pain in the neck.

**WHAT IS THE TREATMENT?**

The methods of treatment for thyroid cancer include surgery, radioactive iodine treatment, external beam radiation therapy and chemotherapy. In most cases, two or more of these methods are used. Surgery is the main treatment for all types of thyroid cancer and is used in almost every case, except for anaplastic thyroid cancer. Usually, a thyroidectomy is done, in which the entire thyroid gland is removed.

**Radioactive Iodine Therapy:** Thyroid cancers take iodine into their cells as part of normal functioning. The Radioactive Iodine (RAI) can be used to kill any remaining cancer cells. This treatment is used to destroy any thyroid tissue which is not removed by surgery and to treat the thyroid cancer that has spread to the lymph nodes and other parts of the body. Radioactive Iodine is usually given as a capsule. Radioactive Iodine therapy is generally not used to treat anaplastic carcinomas. For the therapy to be effective, the patients must have high levels of Thyroid-Stimulating Hormones (TSH) in the



**PREVENTIVE STEPS**

**Thyroid cancer is fairly common** and is usually an indolent disease, tending to remain localised to the thyroid gland for many years.

**As most cases of thyroid cancers** are not associated with any risk factors, there is usually no way to prevent its development. Careful examination of the thyroid and screening for patients at high risk could be considered, though the prevention of thyroid cancer is impossible.

blood. After the surgery, this can be accomplished by stopping the thyroid hormone pills.

**TYPES OF THYROID CANCER**

There are four types of thyroid cancer: **Papillary Thyroid Cancer** grows slowly. **Follicular Thyroid Cancer** is less common. **Medullary Thyroid Cancer** develops in the C cells and is frequently part of a genetic syndrome of Multiple Endocrine Neoplasia (MEN). **Anaplastic Thyroid Cancer** is rare and fast-growing. As this type of cancer grows very quickly, it is more difficult to treat successfully. **Papillary and follicular thyroid cancers** make up 80 to 90 per cent of thyroid cancers. When detected early, these two types can be treated successfully, especially in people younger than the age of 50.



**Dr Kumaresh Krishnamurthy, senior ENT surgeon, Apollo Hospital**

**WHAT DOES THE DOCTOR SAY?**

**A careful physical examination** is done by the doctor, with attention to the size and firmness of the thyroid or any enlarged lymph nodes in the neck. Some laboratory tests are also done to determine the function of the thyroid gland.

**A test to determine the**

aetiology of a thyroid nodule is a nuclear medicine study with radioactive iodine.

**The simplest and often definitive step** in a diagnosis is a Fine Needle Aspiration (FNA). After a diagnosis is made, further tests are done to determine if there is a spread of the disease.

**classact**

## Incubate talent, and then mentor genius

MS Ramaiah Institute of Technology, Bangalore has a full-fledged incubation centre established in June 2004. **Rekha SK** speaks to **Team DNA** on the importance of incubation centres in colleges

Every student has the potential to initiate a start up company, irrespective of the background he/she is from. It is not only the management students who are required to learn how to set up their own companies. A student requires only the willingness and the motivation to learn. Proper guidance from colleges is as important. Most of the students in our campus have started the company with no or minimal investment.

If colleges have incubation centres, students should be provided with facilities like computers, fax, photocopy machines, telephone, and address for correspondence and a mentor to understand and guide students. Having such a set-up will encourage students to avail these facilities after their classes and make use of it.

A mentor is required to guide students and motivate them to do something. Students who already have focus should be guided properly. It becomes very important because the students will then develop an interest in doing something different and unique. The main purpose of an incubation centre in a college is to spread the spirit of self-initiative among students.

The mentors can follow a few steps to make this happen as is done in our campus as well. Initially, awareness programmes must be held. They can be conducted after the regular college hours or so. During such sessions, students can be introduced to the effort which is required for them to start their companies.

After the awareness programme, a few students who show real interest can be hand-picked. Select students can be put into the second/advanced level of training. Here, the students are asked about their areas of interest and where they want to start their companies.

From the level of identifying the right students to motivating, mentoring, idea generation and the like, students should be handled and taken forward until they launch their ventures with the help of mentors.

This incubation also becomes necessary for students to know that they can start their own business enterprises and do not have to worry if there are no jobs around. Apart from this, it also gives students a practical lesson outside of their studies.

**Rekha SK, chief project co-ordinator for the incubation centre, MS Ramaiah Institute of Technology**

# For ICSE students help is not on the line

**With the month-long ICSE and CBSE board exams under way, the CBSE board is providing dedicated exam helplines to deal with stress. ICSE students, however, have no such facilities**

**Jayalakshmi Venugopal, BANGALORE**

While anxious parents of tense CBSE students have access to dedicated exam help-lines, ICSE students have only their teachers or families to turn to, for help. "If I'm upset with my performance in some exam, I wouldn't want to tell my teacher about it, let alone worry my parents. Their expectations get in the way of my openly discussing these things with them," says Geetha R, a class X student.

The CBSE helpline has been operational every year prior to the exams since the last 12 years. But no such measure is in place for students of ICSE schools.

Parents feel that emotional support can be provided at home, but having an academic helpline might be more beneficial. "The ICSE students should also have some kind of a support system when dealing with exam-related stress. As par-



**EXAM BLUES:** Most ICSE schools have in-built mechanisms to help students cope

ents, we can help children sort out emotional issues, but they might need some assistance in other form in academics. And during exams, this can only be provided by a helpline," says Uma R, a parent of a class X student.

Without external support systems, most ICSE schools have an in-built mechanism to help students cope with exam-related problems. "We know how to handle such cases. We have spent six to 10 years with students. So, we know and understand them better than any stranger

managing a helpline. Before exams, we sort out all the problems that students, especially low achievers, might be facing. We even advise parents not to pressurise children during their exams," says Mangal Raj, vice principal, St Germain High School.

School administrations state that the students should voice their need for helplines. "If we get requests from students for helplines, we might check with the ICSE board about it. But so far, no one has expressed such a need," says

Daphne Das, principal, Seventh Day Adventist School.

Many students and parents feel that everyone should have an access to such helplines, as exam-related stress is common among all students. "Why should only a few have access to these helplines during exams? I think it's only fair that there be a helpline for the ICSE students as well," says Supreetha Skanda, a PUC student.

Counsellors are also in great demand during the exam season. The cases start trickling in ahead of exams, from the month of January and peaks during February and March. "I have been seeing a significant rise in the number of exam-related cases, since the last one-and-a-half months. Prior to the board exams, we even had cases of children refusing to sit for their preparatory exams. Now that the exams have started, they (children and parents) will come to us only if they have a problem," says Ali Khwaja, counsellor, Banjara Academy.

School administrations are unsure if these helplines can actually address the real problems efficiently. "I don't think a helpline can make much of a difference to students during exams. The teacher-student rapport might actually help the child sort out issues more successfully. The coping-up mechanism should be built within the education system," says Capt Unnikrishnan, founder-trustee, Primus Public School.

## Group with the power to bring change

**GroupM has gone around the country, working with NGOs and bringing joy to the lives of the underprivileged**

**Team DNA, BANGALORE**

Recently, across the Asia-Pacific region 5,000 team members of GroupM, and its agencies collaborated with various social organisations to make a difference to the lives of people. In India, they worked with underprivileged children, the elderly, and children with special needs.

GroupM also organised a country-wide blood donation drive that saw 500 people participating in the initiative. Across the country, GroupM and its affiliates have been working

tirelessly to make a change. Here are some of their initiatives:

In Mumbai, GroupM has collaborated with NGOs like SHED, Mobile Crèche, All Saints, Shepherd, Andhakshi Ashram, and Urban Community Development.

The objective was to spend some quality time with the needy, provide essential support, not just in terms of time, but also identify the critical needs of the NGO and provide those to them. The team also helped create awareness about the NGOs they visited and committed themselves to garner more public support for them. In addition, there was a blood donation drive, organised at the Peninsula Complex under the aegis of Rotary Club and about 110 employees donated blood.

In Delhi, GroupM worked with two NGOs, Pratham and Action for Autism, where it was involved with slum children and children suf-



**BLOOD BROTHERS:** A GroupM blood donation camp in progress

fering from autism.

It also organised a Blood Donation Camp with the help of the Rotary Club and 40 people came forward to donate.

In Bangalore, two activities took place. One was the visit to an old age home, Little Sisters of the Poor, where they sponsored the breakfast, donated clothes and spent time with the old people who were delighted that an organisation actually took the initiative to come and spend some time with them.

The second activity was the Blood donation camp conducted by the Lions Blood Bank

where 33 people came forward to donate blood. In Chennai, the team visited two NGOs, one was Action Aid that works with abandoned kids, and the other was Vishranti, an old age home.

GroupM spent time at both places, gave away gifts, and also worked towards identifying the key issues and exploring ways in which they could support them on an ongoing basis.

In Kolkata, the team visited an old age home where they spent the day with the residents, helping them do their daily chores, playing games and also gave away medicines.