FSM/AGM CJD Support Network
13th November 2021
MacDonald Spa Hotel
Manchester

Present: Prof Richard Knight, Prof Simon Mead, Andy Tomaso, Bethan Marsh, Lisa Denton, Annette Beal, Margaret Leitch, Brian Marsden, Anita Tipping, Dr. Akin Nihat, Linda Lovett, Gillian & David Turner, Megan Marsh, Lizzie Hill, Morgan Thomas [additional attendees names redacted] (32)

Apologies: Dr. Kate Dahill [additional apologies names redacted] (9)

1.0 Welcome

Prof Knight welcomed everyone for attending the FSM/AGM, informed and re-assured everybody that hand sanitizers and masks are available. Wearing of mask is not mandatory and if one wishes to wear one that will be fine too. This was followed by the health and safety instructions.

The committee members introduced themselves to the members with a short bio of their involvement with the CJDSN.

2.0 "Icebreaker"

This was led by Prof Simon Mead. The members were asked to group themselves per the criteria given like geographically, strain of CJD they are involved in, length of time they have been involved with the Network, members currently caring for a love one and the first-timers attending the event. Moving around and engaging with the other members put everyone at ease before the day's proceedings.

3.0 Approval of the minutes of the virtual 2020 AGM

The minutes were perused and no corrections or omissions were made. Proposed by Annette Beal and seconded by Beth Marsh.

4.0 Matters arising from the 2020 AGM minutes

The list of business matters requiring completion have all been achieved during the year, therefore there are no matters arising.

5.0 Introduction of our New Coordinator

Prof Knight introduced Miss Bethan Rose Marsh as our new coordinator. Beth gave a short summary of her involvement with the Network and her plan to take the Network forward. She was warmly welcomed by the members.

6.0 Chairman's Annual Report

Prof Knight gave a brief overview of the Network's achievements, activities and performance in 2021. In spite of the pandemic caused by Covid-19 and the lockdown, the management committee shared the tasks to keep the Network going. Committee meetings were held on-line via zoom. Four on-line family support meetings were arranged and attended by families. The on-line talks/topics were recorded and are available for CJDSN members to watch. More such meetings are being considered and the situation is under constant review.

7.0 Financial Report

Mr Andy Tomaso, our treasurer spoke of the historical facts on how the Network was funded from its inception to date. Historically, the CJDSN has received funds from the Department of Health (DOH) for £30,000 per annum to develop the helpline, its core values, education and support strategies. The grant ceased in 2010 and the work is able to continue because of the fundraising and donations from families and friends of the CJDSN.

To date, we have the policy to have enough funds to function and run the Network for three years. Surplus allows the Network to give care grants and research grants.

So far x2 research funding had been granted. One research project will be presented today and the 2nd research project is still in progress for completion and will be presented to the members in due course.

The Network is in a stable financial footing to date but still in need of some funds to keep its work going.

Account from 1st April 2021 to 30th November 2021

Total Income	£22412.33
Expenditure	£30904.95
Net Profit	-£8492.62

As of 11th November 2021

Current Account	£4866.37
Reserve Account	£186008.96

8.0 Overview of Prion Disease- Prof Richard Knight

Prof Knight's talk started with basic biology. The body is made up of organs which are made up of cells. Different organs in the human body have different cells that reflect their functions. A cell has a nucleus and it is the basic structure and the functional unit of an organism.

The brain is made up of cells and neurons. Neurons are nerve cells and are electrically active and communicating with each other. The synapse is the conduit between two neurons, the network which transmits information by electrical and chemical signals and this interaction makes the power of the brain. If the synapses malfunction the network breaks down and problems will develop.

Proteins are made in the cell nucleus. Amino-acids are the building blocks of proteins and their functions depend on its final folded phase. Normal body cell contains 46 chromosomes, 23 pairs from each parent.

Each chromosome contains a gene which is a string of code and each code is translated in a chain of amino acids. Mistakes can occur in a string of code. This could be an insertion, a deletion, omission or a change in gene code.

8.1 Important gene code change

1. Pathogenic mutation affects the structure and functions of proteins causing disease.
2. Polymorphism- minor change has occurred but directly not disease causing, however, it may affect one's susceptibility to the disease.

Prion Disease

Fundamentally it relates to the normal prion protein undergoing a change in structure into abnormal protein PrPsc. The human body produces normal prion protein, PrPc. Infection requires an infective agent and the PrPsc is the infective agent where the current view is a misfolded structure.

Incubation could last long and symptoms are progressive, neurons malfunction and die and the disease is untreatable at the moment.

8.2 Types of Prion Diseases

1. Genetic mutations- an autosomal dominant inherited prion disease. This prion disease includes Gertsmann-Straussler-Scheinker (GSS), Fatal Familial Insomia (FFI) and Genetic CJD. The children have a 50/50 chance of contracting the disease.

2. Acquired prion disease- iatrogenic, variant CJD, blood transfusion (secondary transmission), kuru.

3. Sporadic CJD- happens by chance and there are no known clear risks factor. The disease is progressive and life expectancy is around 4-6 months.

8.3 Test that help support the diagnosis

Specific tests 1- find abnormal PrPsc outside the brain, in CSF, blood, skin biopsies, urine and nasal brushings. PrPsc maybe present but at low levels.

Specific test 2- RT Quick detects the abnormal protein if one has the disease. RT Quick is the synthesizing of a normal protein if misfolded protein is found or detected.

All tests need to be evaluated before being put into use.

8.4 Points to consider when making a Clinical Diagnosis

Onset of prion illness is often very non-specific.
Atypical prion disease is difficult to diagnose.
Exclusion of other possible diseases take time.
Passage of time maybe a diagnostic tool.
Emerging specific tests are changing diagnosis.

Post presentation Qs & As followed.

9.0 Treatment: Where are we now?- Prof Simon Mead

PRN 100 treatment for CJD- some of the information about the clinical trial are not ready for public knowledge yet.
PRN100 is the humanized antibody version of ICMS 18, originally shown to be effective against mouse prion disease.

PRN 100 binds to the normal prion protein and prevents the prion seeds from growing.

Through the use of slides, Prof Mead explained how prion seeds grow, and how PRN 100 affects the clearance of PrPsc.

9.1 Active Treatment Strategies

1. Find the drug that binds the normal protein and stops it binding to the prion seed.
2. Find a drug that diminishes the normal prion protein to stop making normal protein (genetic deletion).
3. PRN 100 binds strongly to the normal prion proteins which stops ICMS 18 from seeding.

9.2 Human PRN 100 Immunotherapy

Special needs license was sought and given to treat 6 patients.

Clinical approach on special needs treatment plan was approved by the hospital oversight committee.

Incremental intravenous administration of 1mg/kg, 10mg/kg, 80mg/kg will be given at 48-72 hours interval.

Patients are closely monitored for side effects.

Patients who are at the end of their life won't get this treatment.

9.3 Outcome

Four of the 6 patients have now died.
Two are still alive.
There is no evidence of the drug causing harm.

Qs and As followed after the presentation.

10.0 Predicting Prognosis and Care Milestones in CJD- Dr. Akin Nihat
Dr. Akin Nihat is a neurologist working at the National Prion Clinic.

Slides and statistics were presented showing the changes from the start of the disease, its progression and the predictive life span (4-6 months) of someone diagnosed with sCJD.

The National Prion Monitoring Cohort Data has been gathering data and information for more than 10 years now. The aim of the study is to predict evidence-based milestones. The study aims to collate these data and information and see if any changes have occurred, put them in sequence to tract their progress and pick up key changes that are relevant to the patient's mobility, continence, speech and cognitive function.

Most patients have incomplete data. Collection of information and data as the disease progresses is a tool to aid prediction when outside care is going to be needed.

By using various algorithms, the data and information collected and patterns of changes documented can help predict future data. The result showed 50% of these patients will need care within 30 days and over half of those patients will die within a month.

Future Plan- (1) to collect more data, social care data, use of newer tools/information and biomarkers. (2) Communicate and share the findings to the relevant health care agencies to facilitate a continuity of care.

Post presentation Qs and As followed.

11.0 Replies to submitted questions

Questions submitted by the members were wide ranging and thought provoking, by which all experts in the room took turns in answering them. Some questions took one or two experts to shed light from a different perspective. There were a good number of questions submitted. Few samples below.

1. The effects of the pandemic on seeing patients face to face, is face to face consultation likely to happen or are we moving on to a "zoom" type consultation?
2. Is there any plan for counselling therapy to support the families?
3. Is genetic washing possible before an IVF procedure?
4. Are surgical instruments possible to clean?
5. What is the best way to raise CJD awareness?
6. Neuropsychologist related research??
7. What are your thoughts about assisted suicide?

12.0 Election of Officers

The incumbent committee members are duly re-elected. The election of the officers is accepted and agreed by the members, proposed by Gillian Turner and seconded by Bryan Hobson.

13.0 Care of patients with CJD and their care givers- Margaret Leitch

Care in the hospital- visiting the patient in the hospital has been challenging due to the restrictions imposed by Covid-19. A home visit from the NCJDRSU will be arranged to see the patient and talk to the family. Discharge management and symptoms management and forward planning will be discussed with the family and nurses caring for the patient. The nurses and specialist nurses will be advocates for the patient and the family.

Care at home- a package of care is agreed depending on the availability of staff, specialist nurses and family carers. It involves teams of health care professionals and home helps. Without this group of professionals good care cannot be had or provided.

CHC funding is funding provided to meet the cost of an individual's care in full where it is established that their care need is primarily due to health reasons. Health care checklist is the first stage to be assessed.

Fast care funding- application for this funding is assessed by a multi-disciplinary team. The only requirement is, that the individual has a rapidly deteriorating condition, maybe entering a terminal phase.

CJD Care Package Funding- the funding is mainly for people diagnosed with CJD. The package funding ensures that shortfalls in the care provision are met and payment can only be done through the CHC.

A diagnosis of CJD on a family member will change the life of the family/carer for an unforeseeable future. Families/carer with the right support will find new ways of doing things, will reach out to friends for support when things get overwhelming to cope with. Carers are entitled to have their needs assessed and they too need respite from caring by getting a paid carer. There is no one template on how to cope with the stress while caring for someone with CJD. Carers are advised to slow down, have a downtime for reflection to help manage their expectations.

Benefits for carers- carers are eligible for one or more state benefits to help with the cost and potential loss of earning and they are eligible for a carer's allowance.

Qs and As followed post presentation.

14.0 Round Table Discussion

Members/attendees randomly joined the round table discussions. The

experts and the committee members joined each table for advice and further support.

15.0 Raffle Draw

Prizes are displayed for the members to see. Winning tickets holders who cannot claim the prize on the day will have their prizes posted to them.

16.0 Close

Prof Knight summarized the day's proceedings and thank everyone for attending the Family Support meeting, for their continuing support and wished everyone a safe journey home.

The FSM/AGM finished at 1600hrs.