



## FAMILIES *for* FAMILIES DAY

Saturday 13<sup>th</sup> May 2017

THE HILTON BIRMINGHAM METROPOLE HOTEL  
NATIONAL EXHIBITION CENTRE, BIRMINGHAM B40 1PP

### *Programme*

**09.15 The Registration Desk opens and refreshments available**

**10.00 Chairman's Welcome and Who's Going Where?**

Ian Legg, NSA Chair, welcomes families and introduces the day; and it's time to decide which break-out groups the young people and children will join. For everyone else, stay in the Main Room for the main programme:

**10.10 The Annual General Meeting of the NSA charity**

Starting with the AGM, a short open meeting, at which the various positions on the charity board are confirmed for the coming year, Ian Legg sums up progress over the last 12 months for the charity and looks ahead.

**10.25 The Chairman introduces the Medical Advisory Group**

**10.30 The CASPER Study**

Dr Shruti Garg reports on the official outcomes of the CASPER Study (Cognitive and Social Profiling: Exploring Rasopathies) and the practical implications for families

**10.50 GenIDA Patient Registry**

Dr Florent Colin introduces this ground breaking development which you can be a part of.

**11.20 The Medical Panel and guests – Questions & Answers**

**Professor Michael Patton, Consultant Clinical Geneticist & our Medical Adviser** chairs a Q&A session when you can ask your questions to our team of medical experts from the NSA Medical Advisory Group. Our Panel:

**Professor Bronwyn Kerr** - Consultant Clinical Geneticist, St Mary's Hospital, Manchester

**Dr Juan Pablo Kaski** - Consultant Paediatric Cardiologist, Great Ormond Street Hospital

**Dr Pia Ostergaard** - Senior Lecturer in Human Genetics, St George's Hospital

**Dr Shruti Garg** - Clinical Senior Lecturer in Translational Child Psychiatry, University of Manchester

**Dr Kostas Dimopoulos** - Consultant Cardiologist in adult congenital heart disease & pulmonary hypertension,  
Royal Brompton Hospital

**Dr Vin Diwakar** - Regional Medical Director, NHS England London

**Judith Van De Meerakker** - Department of DNA Diagnostics, Amsterdam Academic Medical Centre

**Dr Florent Colin** - GenIDA project manager

**Linda Shaughnessy** - Clinical Nurse specialising in adult congenital heart disease, Royal Brompton Hospital

**12.40 Fundraising/"Reach Out For Noonans"**

Ian Legg briefly explains the Regular Giving Scheme and how the wider family and friends can help.

**12.45 Lunch**

An informal buffet giving you more opportunities to talk to the guest specialists and other families, learn more and register with the GenIDA project and become a regular giver to NSA.

**Support for individuals and families affected by Noonan Syndrome  
and related conditions (on the Ras/MAPK pathway) in the UK**

Registered Charity No. 1140671

Medical Adviser: Professor Michael A Patton FRCP

### **13.45 Discussion Groups**

Delegates gather in their pre-chosen groups:

- **Speech & Feeding**
- **Education**
- **Adulthood**

### **14.30 Refreshments available**

*And another opportunity to talk to the specialists and other families*

### **14.45 The Quest For Finding More Genes For Noonan Syndrome**

Dr Pia Ostergaard and her team from St George's Hospital update us on the latest developments

### **15.15 Potential Future NSA Funded Research**

Professor Michael Patton, Professor Bronwyn Kerr and Ian Legg discuss what areas might be addressed by future research

### **15.30 The Break Out Group Presents...!**

### **15.45 The important messages from the day, closing remarks and farewell**

Ian Legg sums up the day and looks to the future...

**Day Closes by 16.00 at the latest**

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Medical Adviser: Professor Michael A Patton FRCP

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