

Chromosome 2, 4, 7, 15

Reference Number: RDF1052-22

Date of Response: 01/12/22

Further to your Freedom of Information Act request, please find the Trust's response(s) below:

Dear Royal Devon University Healthcare NHS Foundation Trust,

Please provide the following data.

- 1) *A list of all recognised medical conditions caused by microdeletions and microduplications on chromosome 2, chromosome 4, chromosome 7, chromosome 15.*

Answer: The Trust does not hold this information.

- 2) *A table of symptoms which are found in patients with microdeletions and microduplications on chromosome 2, chromosome 4, chromosome 7, chromosome 15.*

Answer: The Trust does not hold this information.

- 3) *How many patients in the UK (or the Trust, if data isn't held for the UK) have microdeletions or microduplications on chromosome 2, chromosome 4, chromosome 7, chromosome 15?*

Answer: The Trust does not hold this information.

- 4) *Of the patients in part 3), how many have recognised medical conditions caused as a result of the chromosomal issue?*

Answer: The Trust does not hold this information.

- 5) *Of the patients in part 3), how many have symptoms caused as a result of the chromosomal issue?*

Answer: The Trust does not hold this information.

- 6) *Please provide a table of the symptoms presented by patients in part 5), along with a percentage indicating the prevalence of these symptoms, i.e. Growth Issues - 60%.*

Answer: The Trust does not hold this information.

- 7) *Expressed as a percentage, how common are the following symptoms in patients with chromosomal abnormalities on chromosome 2, chromosome 4, chromosome 7, chromosome 15 (please use a column table to show Symptom, Number of Patients, Percentage of Patients).*

- a) *Gastroesophageal reflux;*
- b) *Feeding Difficulties (Infancy and Ongoing);*
- c) *Failure To Thrive;*
- d) *Frontal Bossing;*
- e) *Wide anterior fontanelle;*

- f) *Hypertelorism;*
- g) *Epicanthal folds;*
- h) *Depressed nasal bridge;*
- i) *Bilateral Cryptorchidism;*
- j) *Micrognathia (small lower jaw);*
- k) *Low-set ears;*
- l) *Posteriorly rotated ears;*
- m) *High arched palate;*
- n) *Hypotonia;*
- o) *Joint laxity/hypermobility in all joints;*
- p) *Gross Motor Delays;*
- q) *Global developmental delay;*
- r) *Unusually happy disposition;*
- s) *Autism Spectrum Disorder;*
- t) *Speech and Language Delays.*

Answer: The Trust does not hold this information.