

Lowe Syndrome – A&E Information Sheet

Parents – please complete this and take along with a copy of the most recent clinical report regarding your child.

Patient name:	DOB:
Specialist name:	Institution:
Telephone:	Email:

Clinicians – this sheet provides background information for Lowe Syndrome, also known as Oculocerebrorenal syndrome of Lowe (OCRL). For further information, please use the details above to contact the specialist responsible for this patient, who is happy to advise on details of the medical management.

- Low Syndrome**
- Rare, X-linked recessive disorder, affecting males
 - Causes physical and mental handicaps
 - Affects the eyes, brain, kidneys, muscles and bones
 - There is currently no cure, and treatment is supportive



- Eyes**
- Born with cataracts, usually removed early in life
 - 50% of cases suffer raised intra-ocular pressure leading to glaucoma



- Kidneys**
- Kidney disorders can cause features of renal Fanconi syndrome, including:
 - o polyuria and/or polydipsia; this can lead to dehydration
 - o low-molecular weight proteinuria
 - o elevated urinary calcium, which can cause kidney stones
 - o metabolic acidosis
 - o phosphate wasting, which can cause rickets



- Abdomen**
- Pain can be due to constipation resulting from dehydration
 - Acute pain can also be caused by kidneys stones, so an U/S should be considered



- Surgery/ blood**
- If surgery is required that risks major blood loss, it is important to note that many Lowe's patients have an impaired platelet function, evident from prolonged closure times in the PFA-100 system. The bleeding risk can be ameliorated with ϵ -aminocaproic acid.

