



*I wish to  
have a white  
fluffy puppy*

**Jade, 7**  
genetic disorder



# Genetics Referral Guidance Sheet

*To qualify for a wish, a child must meet these criteria at the time of referral:*

1. Older than 2½ years and younger than 18 years
2. Has not received a wish from another wish-granting organization
3. Diagnosed with a **critical illness** (i.e. a progressive, degenerative or malignant condition) that, despite adherence to the treatment plan, is currently placing the child's life in jeopardy

## Qualifying Genetics Conditions:

- ✧ Aicardi Goutieres disease
- ✧ Alexander disease (symptomatic)
- ✧ Barth syndrome
- ✧ Congenital anomaly, chromosomal or single-gene condition with associated life-threatening complications such as:
  - Associated major gastrointestinal dysfunction meeting gastroenterology criteria
  - Heart anomalies meeting cardiology criteria
  - Kidney dysfunction meeting nephrology criteria
  - Motor seizures meeting neurology criteria
  - Respiratory complications meeting pulmonary criteria
- ✧ Gaucher disease, type 2
- ✧ Infantile Pompe
- ✧ Krabbe disease
- ✧ Leigh disease with MRI changes
- ✧ Lesch-Nyhan syndrome
- ✧ Maple syrup urine disease with a history of hyperleucemia event occurring after diagnosis
- ✧ Metachromatic leukodystrophy, progressive
- ✧ Mucopolysaccharidosis disorders (symptomatic) such as:
  - Hunter syndrome
  - Hurler syndrome
  - Sanfilippo syndrome
- ✧ Niemann-Pick disease
- ✧ Peroxisomal disorder
- ✧ Pyruvate dehydrogenase deficiency with progression of disease
- ✧ Skeletal dysplasias or dysostosis with progressive pulmonary complications requiring additional supportive measures (i.e., BiPAP/CPAP or chronic ventilator dependence)
- ✧ Sphingolipidosis with symptoms in the pediatric period:
  - GM1 gangliosidosis
  - Tay-Sachs
- ✧ Trisomy 13
- ✧ Trisomy 18
- ✧ Urea cycle (CPS, OTC, citrullinemia) and organic acidemia disorders (methylmalonic and propionic) with a history of hyperammonemic event occurring after diagnosis
- ✧ X-linked adrenal leukodystrophy with adrenal or brain findings

There are other conditions that may be eligible for a wish when the condition includes life-threatening comorbidities that are currently placing the child's life in jeopardy. These conditions will be reviewed on a case-by-case basis. Please include detailed information on these conditions when submitting the Diagnosis Verification Form.