Social Security National Disability Forum
Compassionate Allowances and Rare Diseases

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MLD Foundation

- Co-Founded with my husband in 2001
- Hosted and chaired conferences in Melbourne, Tokyo, Munich, London, etc., and all across the US … for 18 years
- Met and collaborated with dozens of researchers from around the globe … mice, natural history therapy development, quality of life, disease burden, etc.
- Primary “go to” for MLD information for at least 5 pharmaceutical companies working on MLD
- Very active in MLD and at the Rare Disease level: FDA-PFFD, NIH-NINDS, policy (newborn screening - RUSP Roundtable, Rare.Army), registries, etc.

facilitating **C**ompassion
increasing **A**wareness
influencing **R**esearch
promoting **E**ducation

... for Metachromatic Leukodystrophy

Personal Connection to MLD …

Darcee …
Passed away at age 10, ≈22 years ago.

Experimental early stage bone marrow transplant – MN/Krivit

Lindy …
Diagnosed at 14 (1995) after a 6 year diagnostic odyssey

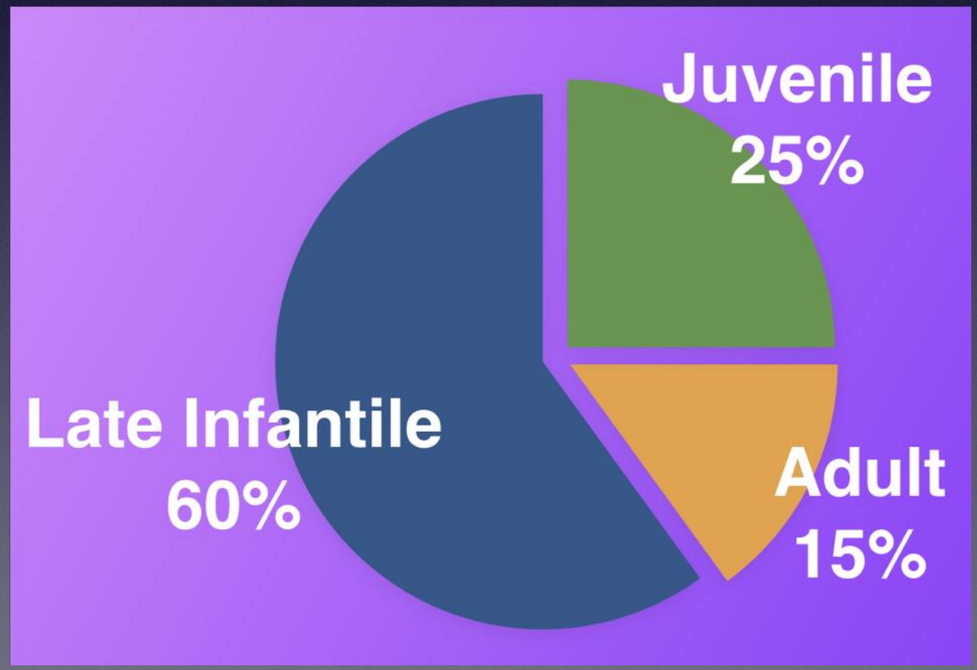
Docs told us 4-6 yrs.

She’s 37 now and doing much better than we/they anticipated

- MLD/Rare Disease mom
- We personally know nearly all of these MLD families
- Former pediatric RN

Metachromatic Leukodystrophy

- Very rare terminal genetic neuro-metabolic disease where over half the cases affect infants. (Late Infantile MLD)
- Juvenile and adult forms too
- Only Late Infantile has a Compassionate Allowance



Late Infantile vs Juvenile MLD

- Impairment Summary - DI 23022.235 Late Infantile MLD
- Juvenile Form Differences
  - Begins between age 4 to 16
  - Can start with motor or cognitive difficulties
  - Mis-diagnosed, diagnostic odyssey lasting years
  - Same diagnostic testing used

Incidence & Prevalence

1:40,000 births has MLD

	Annual Births			Alive		
	USA	More Developed Countries	Global	USA	More Developed Countries	Global
Late Infantile	63	191	1,996	250	760	8,000
Juvenile	28	87	907	540	1,650	3,600
Adult	23	69	728	1,120	3,500	37,000
Total	**114**	**347**	**3,630**	**1,900**	**6,000**	**48,700**

Source: MLD Foundation

Pseudo Deficiency

- Occurrence is I in 12 in population
- ARSA A lower than normal
- No signs or symptoms
- **DO NOT excrete sulfatides in urine**
- **Do not have MLD**

MLD’s Classification

Lysosomal Disease & Leukodystrophy

- **Lysosomal Disease … result from defects in lysosomal function**
  - **Abnormal build-up** of various toxic materials in the body's cells as a result of enzyme deficiencies affecting different parts of the body, including the skeleton, brain, skin, heart, and central nervous system
  - **Incidence** of over **50 lysosomal diseases** estimated at **1:5,000**

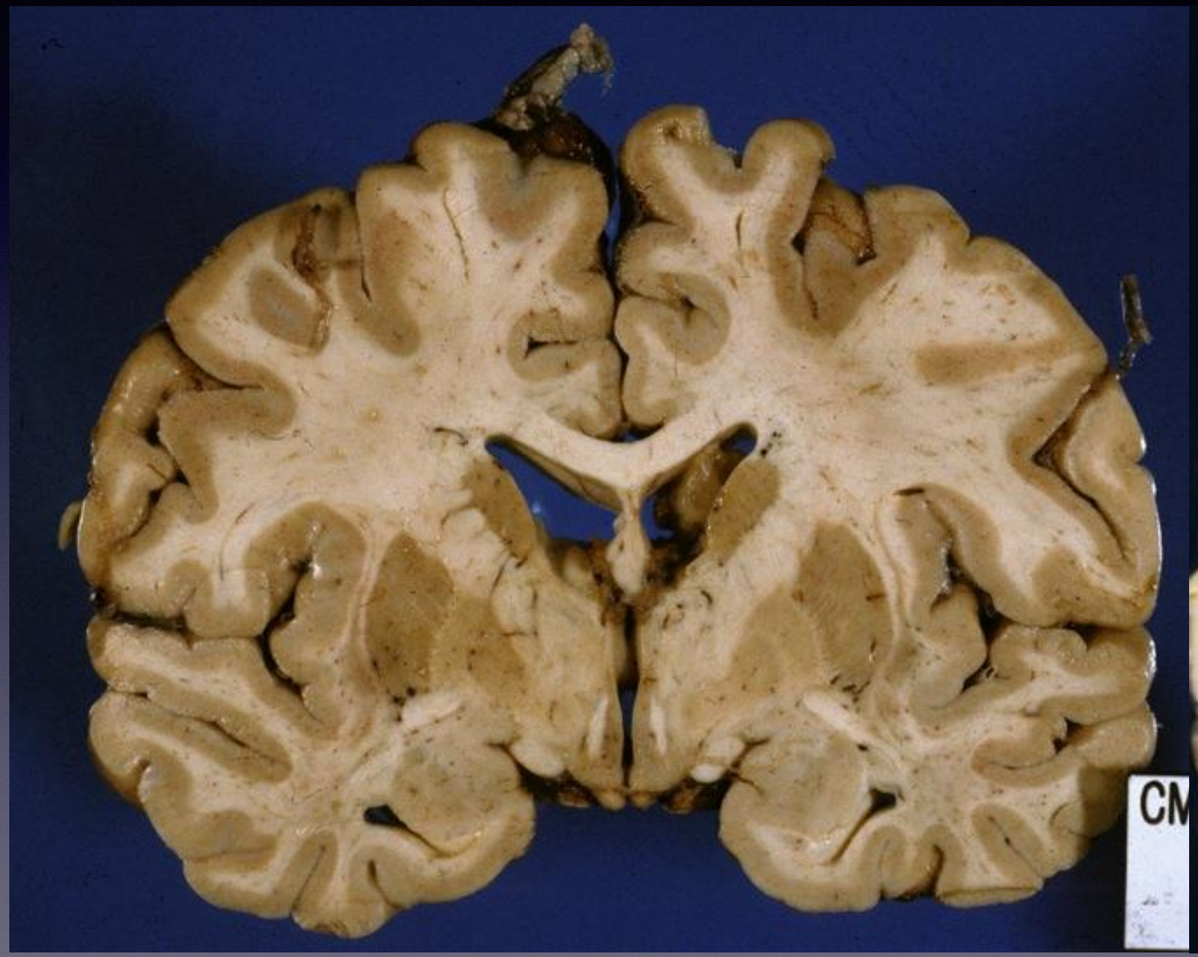
MLD’s Classification

- **Leukodystrophy**
  - **Abnormal development or destruction** of the white matter (myelin sheath) leading to a range of central and peripheral neurological problems
  - **Incidence** of over **50 leukodystrophies** is estimated at **1:7,600**

Testing/Diagnosis

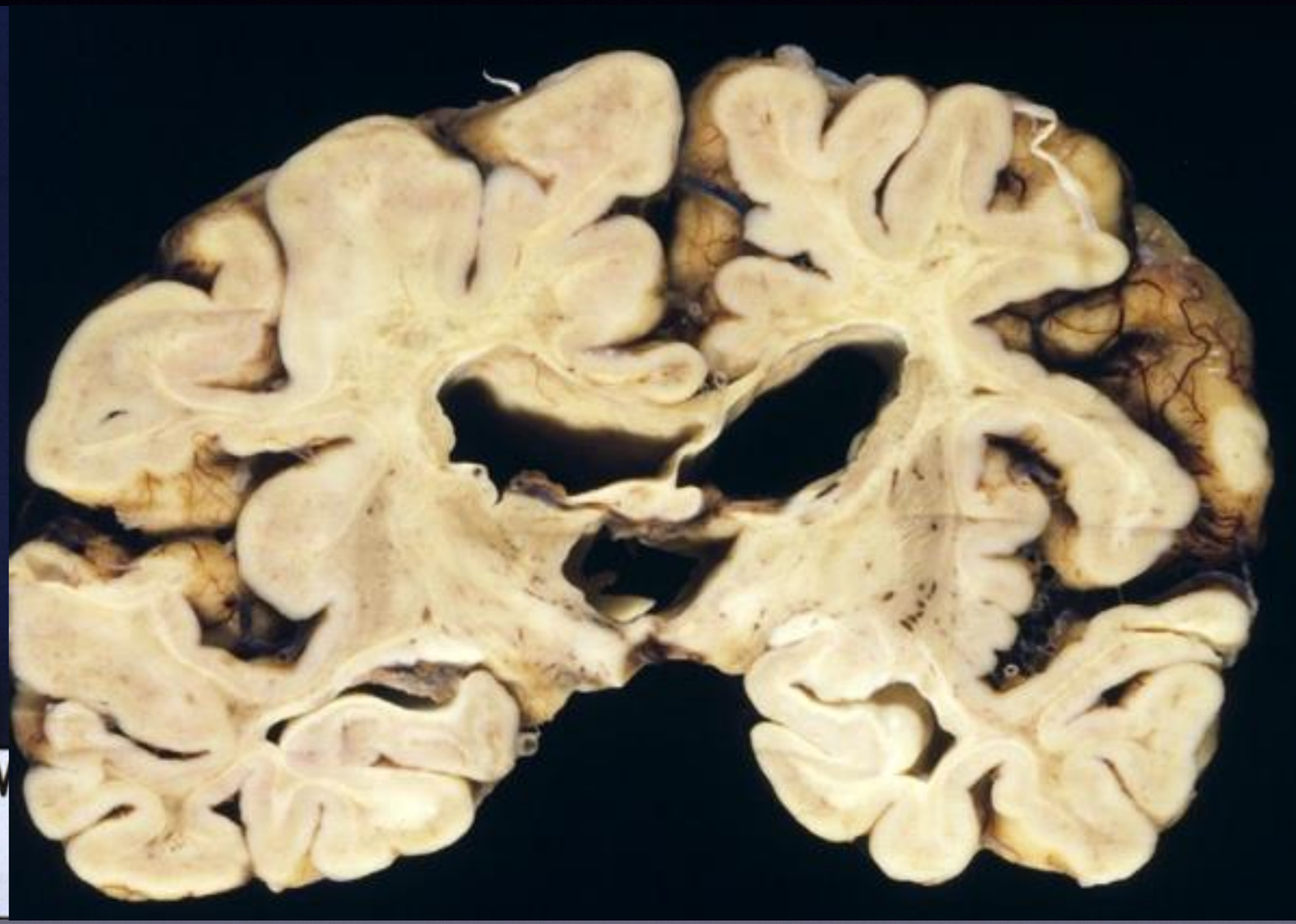
- Must rule out pseudo deficiency by confirming low ARSA levels plus secreting sulfatides in urine or by genetic testing
- MRI helpful - identify MLD lesions and atrophy in white matter of brain characteristic for MLD, but not confirmatory
- ICD-10-CM code E75.25
- Working on Newborn Screen for MLD detecting sulfatides in the blood

Normal White Matter



http://cclcm.ccf.org/vm/VM_cases/neuro/neuro_case1_gross1.jpg

Loss of White Matter



http://www.neuropathologyweb.org/test6/6testimages/6p22G-XALD.jpg

Juvenile MLD

- Is genetic … it will last over 12 months … and is terminal
- Is usually diagnosed after symptoms appear
- Results in severe cognitive and motor skill challenges
- Will never have gainful employment
- Will always require assistance and supervision

Frankly, it's no different than infantile MLD other than age of onset

Juvenile MLD

Therapy … stem cell transplant if very early symptomatic

- Children lose additional capabilities during therapy
- Nearly always wheelchair bound after transplant
- They will continue to progress from this lower functional level .. but at a slower rate

So even those with therapy become or remain disabled

Adult MLD

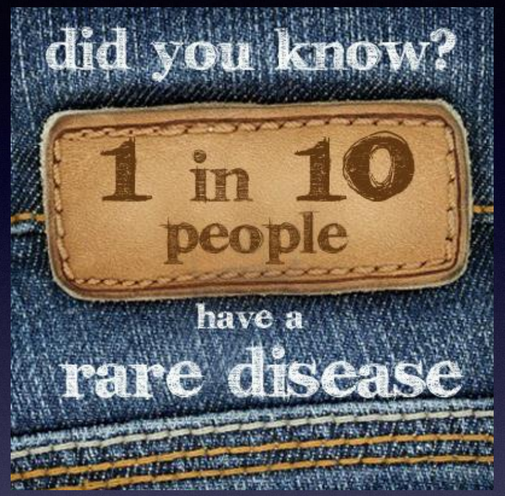
- Is genetic … it will last over 12 months … and is terminal
- Is usually diagnosed after symptoms appear .. cannot function at work
- Results in severe cognitive and motor skill challenges
- Will quickly lose gainful employment skills … sequencing, memory, impulse control, bowels, etc.
- Will soon require assistance and supervision … and then a care home

Frankly, it's no different than infantile MLD other than age of onset

Forum Purpose

- Focus on learning additional information about the rare disease community
- Identify conditions we should consider adding to the List of Compassionate Allowances (CAL)

Rare Disease Facts



Source: **Global Genes Rare Disease Facts** https://globalgenes.org/rare-diseases-facts-statistics/

Rare Diseases are not always terminal or disabling …

Rare Disease

- **But when they are …**
  - the impact is tremendous
  - often increase medical needs & expenses
  - quality of life and practical care needs & expenses
  - loss of ability to work … 2 incomes may become 1 .. or none!

Rare Disease

- **Worse yet …**
  - Rare diseases are often under or misdiagnosed
  - and confirmed only after a long diagnostic odyssey
    - confirmatory diagnostics are often easy and precise … once you know what you are confirming
- When a family **finally applies to SSA for SSI (or SSDI) they are worn out and often desperate**

SSI

- CAL is just the first step
- Still need to meet Financial eligibility
- But even a denial can open the doors to Medicaid Waivers

Discussion Points from Survey

- CAL - Best kept secret
  40% didn’t know what it was, 100% weren’t told

- Time to Decision - All over the map (less than a month to 3 years)
  Most 2 to 9 months

- Inconsistencies - decisions seem to be dependent on reviewer
  *19 yr old - counted parents income resulting in denial
  *Own car - won’t qualify
  *Own house - won’t qualify, move on base - will qualify

- 18 yr old - 2 denials - had to be 18 for a month to qualify as 18
  had to cash out savings bonds, spend down assets to under $2000 to qualify

- Approved - receiving SSI for 6 month - got bill owed $2000, told it was a mistake but bills continued, finally requested to stop receiving benefits

Process

- Too complicated
- Needed Advocate to apply - lots of man hours
- Needed help from Hospital
- Could be approved but benefit would be $1/day and would go away if admitted to hospital due to insurance involvement - advised to take denial.
- Not told about differences between SSI and SSDI - would qualify for SSI
- 800 number on hold forever - went to local office

Appeals

- Approved after 4 appeals - took 9 months - too much red tape
- 3 appeals - took 3 years - said didn’t meet disability eligibility despite terminal condition, unable to work

Open Discussion

- Fighting to be on CAL but need to open discussion around creating a better less complex system and revisiting financial eligibility requirements in the face of the complexities of Rare Disease

- If we are successful here in improving this system, next steps would be to open discussion on how a program like CAL might help Rare Disease families obtain Medicaid sooner.

Summary

- Rare Diseases affect 1 in 10 Americans
- MLD is a rare genetic terminal disease that affects cognitive and motor skills
- Late infantile MLD is already on the CAL
- Juvenile and Adult MLD are almost identical to late infantile MLD from a disability respective … only the age and size of the patient is different
- **We are requesting juvenile and adult MLD be granted a CAL**

Thank You!