



PREVENTION OPPORTUNITIES UNDER THE BIG SKY

Cystic Fibrosis Awareness Month in Montana

Cystic Fibrosis (CF) is a genetic disease which affects the respiratory and digestive systems.¹ Common manifestations of this disease include persistent pulmonary infections, pancreatic insufficiency, and elevated sweat chloride levels. In some circumstances, an individual may demonstrate mild or atypical symptoms, and clinicians should remain alert to the possibility of CF even when few of the usual symptoms are present.²

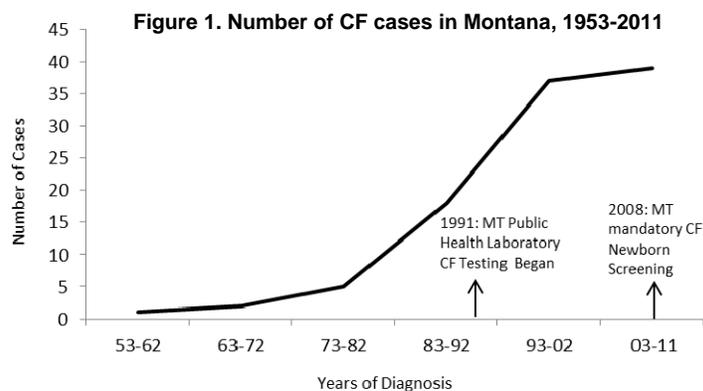
The life expectancy for persons with CF has increased dramatically from about 25 years during the 1980's to about 37 years at the current time. Timely diagnosis and treatment can greatly improve the lifespan and quality of life for person with this disease.² In 2008, CF was added to the mandatory newborn screening panel in Montana to help identify infants who would benefit from early diagnostic testing and treatment. This issue of *Montana Public Health* presents findings from the CF Foundation Patient Registry for CF individuals born before 2008, describes CF signs and symptoms, recognition and treatment, and recommendations for health care providers.

Epidemiology of CF Nationally, approximately 30,000 Americans have CF and an estimated 1,000 new cases are diagnosed each year. This disease is most prevalent in Caucasians (1:2,500).²

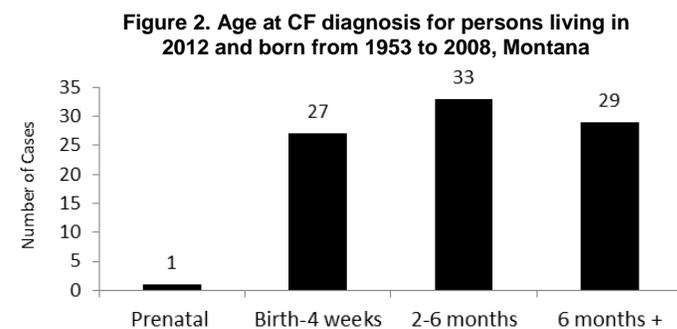
CF in Montana An estimated 184 individuals live with this disease in Montana.³ Since the 2008 implementation of mandatory CF Newborn Screening, 12 cases have been confirmed among Montana newborns.⁴ Over the past 50 years, the number of identified CF cases has increased (Figure 1). The steady rise in number of CF diagnoses is possibly due to the recognition of individuals with a mild form of this disease as well as more recent implementation of CF mandatory newborn screening. CF is usually considered a pediatric disease, however Montana has seen an increasing number of late CF diagnoses among adults.

CF in Montana Prior to 2008 The CF Foundation Patient Registry lists 102 patients who receive care at Montana CF Team Clinics. Of those 90 were born prior to 2008. Those individuals had clinical diagnosis prompted by acute or persistent respiratory abnormalities, meconium ileus, failure to thrive, newborn screening, family history, and other symptoms (Table, page 2).

Among the 90 individuals born prior to 2008, one person was diagnosed prenatally, 27 were diagnosed within the first 4 weeks of life and 33 were diagnosed by age 6 months (Figure 2). Almost one-third (29) of these persons were not diagnosed until later in life. The median age of diagnosis among persons with CF was 6 years for those diagnosed at age 6 months or later. The oldest person was diagnosed at age 16 years.



Source: Cystic Fibrosis Foundation Patient Registry, 2011



Source: Cystic Fibrosis Foundation Patient Registry, 2011

Time frames are based on *The CF Diagnostic Process for Screened Newborns*.⁵

Treatment Although there is no cure for cystic fibrosis, timely treatment through medications, therapy and surgical procedures can ease symptoms and reduce complications. The Montana CF Foundation-accredited care center in Billings offers expert consultation regarding diagnosis and treatment (see page 2, *Recommendations for Health Care Providers*).

Table. Signs, symptoms and history that prompted clinical diagnosis of CF, Montana, cases alive in 2012 and born prior to 2008

Finding that prompted diagnosis	Age at diagnosis*	
	Prenatal-6 months	6 months and older
Meconium ileus/other intestinal obstruction	17	2
Acute or persistent respiratory abnormalities	16	18
Failure to thrive/malnutrition	16	12
Newborn (neonatal) screening	16	
Family history	10	3
Steatorrhea/abnormal stools/malabsorption	10	4
Electrolyte imbalance	6	4
DNA analysis	4	
Prenatal screening: chorionic villus sampling, amnio	5	
Edema/ Liver problems/ Rectal prolapse	3	

*Indicates number of patients. Patients may have more than one symptom/condition or test before diagnosis.

Source: Cystic Fibrosis Foundation Patient Registry, 2011.

Recommendations for health care providers

- **Maintain index of suspicion:** Newborn screening may miss up to 5% of cases. Please refer anyone for whom you suspect CF, even if they have had a negative screening result.
- **Recognize conditions common in CF:** Pancreatic insufficiency, chronic pancreatitis, and failure to thrive are common but not always present. Other suggestive conditions include:
 - chronic, progressive respiratory symptoms that do not respond to treatment
 - persistent gastrointestinal symptoms such as severe constipation, blockage, or fat malabsorption
 - rectal prolapse
 - male infertility
- **Prepare for testing referral:** Before sending a patient for sweat testing, please contact the Montana CF Center at 406-235-6614 for referral to an appropriate testing location.

For more information, contact MT CF Center in Billings: Jerimiah Lysinger, MD or Rosalie Bush, RN, MSN: (406) 235-6614, MT Newborn Screening Program:(800) 821-7284, MT Children's Special Health Services (406) 444-0043.

References:

1. Ratjen F. *Cystic Fibrosis*. Lancet. 2003; 361:681.
2. American Lung Association. *State of Lung Disease in Diverse Communities*.2010:41.
3. Child Health Referral Information System (CHRIS), Montana, 2011.
4. Newborn Screening Program, Montana, 2011.
5. Farrell P. Guidelines for Diagnosis of Cystic Fibrosis in Newborns through Older Adults: Cystic Fibrosis Foundation Consensus Report. *J Pediatrics*, 2008; 153: S4-S14.

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