



# Disability Awareness Begins With You: ATR-X SYNDROME

Alpha thalassemia X-linked mental retardation (ATR-X) syndrome is a rare genetic disorder affecting multiple organ systems of the body. ATR-X syndrome is characterized by mental retardation, characteristic facial features, abnormalities of the genitourinary tract, and alpha thalassemia. Alpha thalassemia, which is a condition where there is a defect in the production of the oxygen-carrying pigments of red blood cells (hemoglobin), is not seen in every case. Additional abnormalities are usually present in most cases. ATR-X syndrome is inherited as an X-linked recessive trait.

Some researchers have suggested the name XLMR-hypotonic facies syndrome be used to designate several disorders formerly considered separate entities including ATR-X syndrome, Carpenter-Waziri syndrome, Chudley-Lowry syndrome and Holmes-Gang syndrome. All of these syndromes occur due to mutations of the same gene on the X chromosome. Some researchers prefer use of the name ATR-X syndrome because it is the most widely-recognized term for this disorder.

## Synonyms of Alpha Thalassemia X-linked Mental Retardation Syndrome

- ATR-X Syndrome
- Carpenter-Waziri Syndrome
- Cerebrofaciogenital Syndrome
- Chudley-Lowry Syndrome
- Holmes-Gang Syndrome
- XLMR-Hypotonic Face Syndrome

## Organizations related to Alpha Thalassemia X-linked Mental Retardation Syndrome

March of Dimes Birth Defects Foundation  
1275 Mamaroneck Avenue  
White Plains, NY 10605  
Phone #: 914-428-7100  
888-663-4637

e-mail: [skus@marchofdimes.com](mailto:skus@marchofdimes.com)  
<http://www.marchofdimes.com>

NIH/National Institute of Mental Health  
6001 Executive Blvd  
Rockville MD 20892-9663  
Phone #: 301-443-4513  
e-mail: [nimhinfo@nih.gov](mailto:nimhinfo@nih.gov)  
<http://www.nimh.nih.gov/>

National Institute of Neurological Disorders and Stroke (NINDS)  
31 Center Drive  
Bethesda MD 20892-2540  
Phone #: 301-496-5751  
800-352-9424  
e-mail: [braininfo@ninds.nih.gov](mailto:braininfo@ninds.nih.gov)  
<http://www.ninds.nih.gov/>

The Arc (a national organization on mental retardation)  
1010 Wayne Ave,  
Silver Spring MD 20910  
301-565-3842800  
800-433-5255  
e-mail: [info@thearc.org](mailto:info@thearc.org)  
<http://www.thearc.org/>

[http://www.rarediseases.org/search/rdbdetail\\_abstract.html?disname=Alpha%20Thalassemia%20X-linked%20Mental%20Retardation%20Syndrome](http://www.rarediseases.org/search/rdbdetail_abstract.html?disname=Alpha%20Thalassemia%20X-linked%20Mental%20Retardation%20Syndrome)