Adrenoleukodystrophy is caused by mutations in the ABCD1 gene, which is located on the X chromosome. As it is an X-linked disorder, it mainly affects males. Females can be carriers of the gene mutation but are usually asymptomatic.

Biochemical Basis: The ABCD1 gene encodes a protein involved in the transport of very long-chain fatty acids (VLCFAs) into the peroxisomes, where they are broken down. Mutations in this gene lead to the accumulation of VLCFAs, particularly in the adrenal glands and the white matter of the nervous system.

Clinical Manifestations: The symptoms of ALD can vary widely, and there are different forms of the disorder. The most severe form is the childhood cerebral form, which typically presents between the ages of 4 and 10. Symptoms include behavioral changes, cognitive decline, loss of muscle control, and visual and hearing problems. Adrenal insufficiency (Addison's disease) may also occur.

Adrenal Gland Involvement: In addition to neurological symptoms, ALD often affects the adrenal glands, leading to adrenal insufficiency. This can result in fatigue, weakness, weight loss, and other symptoms related to hormonal imbalances.

Diagnostic Methods: Diagnosis is often based on clinical symptoms, family history, and biochemical testing for elevated levels of VLCFAs. Genetic testing can confirm the presence of mutations in the ABCD1

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